MINISTRY OF HIGHER AND SECONDARY SPECIALIZED EDUCATION OF THE REPUBLIC OF UZBEKISTAN

MINISTRY OF PUBLIC HEALTH OF THE REPUBLIC OF UZBEKISTAN

BUKHARA STATE MEDICAL INSTITUTE

DEPARTMENT OF CHILDREN'S NEUROLOGY AND PROPEDYTICS OF CHILDREN'S DISEASES

EDUCATIONAL-METHODICAL COMPLEX

for third year students of the Faculty of Pediatrics and Medical Pedagogy on the subject of Medical Genetics

Field of study: 500000 – Public Health and Social Welfare

Field of study: 510000 – Public Health

Field of study: 5510200 - Pediatrics

Bukhara – 2021

The educational-methodical complex of the subject was approved by the statement of the meeting of the Coordinating Council of educational-methodical associations of higher and secondary specialized, vocational education of 2021 "26.VI" _____ "_____".

The educational-methodical complex of the subject was developed at Bukhara State Medical Institute.

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Educational-methodical complex of the subject has been recommended by the Scientific-Methodical Council of Bukhara State Medical Institute. (statement No. “____” in 2021 “___” _____)

CONTENTS:

The subject program is based on the State Educational Standard of the Republic of Uzbekistan and the qualification requirements of the bachelor's degree program and is designed to teach students of pediatric, medical and pedagogical faculties of medical universities. The program determines the amount of theoretical knowledge and practical skills that students should know in the science of nervous system diseases. On the basis of this program, using modern pedagogical technologies in the teaching process, the student learns to perform basic practical skills on phantoms from theoretical knowledge and combine the acquired skills with clinical practice through modern medical technologies concerning the diseases, their diagnostic principles, comparative diagnostic aspects, as well as the basics of prevention (general and special).

Modern achievements in science, engineering and technology in the field, depending on the requirements of personnel customers, there may be additions and changes in the professional activities of bachelors.

The subject of medical genetics is a general professional subject and is taught in 5-6 semesters at the faculties of medicine and medical pedagogy. Adequate knowledge of mathematical and natural sciences (medical biology and genetics, human anatomy, biophysics, normal physiology, bioorganic, bioorganic and biological chemistry) as well as clinical sciences and epidemiology of quarantine diseases, microbiology, virology and immunology planned in the curriculum for the implementation of the program and skills are required. The subject concludes final test control.

LECTURE №1:

History of the development of genetics. Classification of hereditary diseases, etiopathogenesis. Chromosomal diseases (general characteristics). Screening program

1.1. A model of educational technology.

Class time - 2 hours	Number of students: 18
Class type	Lecture
Plan	History of the development of genetics. Classification of hereditary diseases, etiopathogenesis, mutation process, clinic. Chromosome diseases. Skinning software. Frequency and prevention of congenital and hereditary pathologies. Autosomal and sex chromosomal diseases, diagnostic criteria and methods of treatment.
The purpose of the topic: - During clinical practice, the doctor should be able to personally examine the patient, talk to him and provide communicative assistance. During this period, the student must live the life and mood of the clinic. Examines the patient and evaluates the internal and external organs on the basis of the propaedeutics of internal medicine. Assess the patient's condition and help improve his or her condition. The student should behave like a doctor next to the patient.	
Task of the topic - Use of basic genetic testing methods; Conducting treatment and prevention of patients in outpatient and home settings after genetic disease	
Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2. Lecture Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment 5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They are divided into small groups They watch They participate They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

The term genetics was coined in 1906 by U. Betson and is derived from the Greek word “gepeiko5” and refers to its origin. The science of genetics studies the two main characteristics of living organisms - the laws of heredity and variability. G. Mendel began his scientific study of the phenomena of heredity and variability with his classical experiments on points (1865). The laws of heredity discovered by Mendel in Beijing did not attract the attention of contemporaries for 35 years, but from 1900 onwards, after the laws of heredity were discovered, Mendel's laws were recognized by all scientists and genetics began to develop as a separate science. Since then, the science of genetics has come a long and complicated way and has been divided into numerous independent sections. These departments are developing as separate disciplines, formed as a result of the use of new discoveries in the science of genetics - mathematics, physics, chemistry, evolutionary theory, cytology, medicine and other sciences. Genetics is a fundamental and applied science. Its fundamentalism - the study of heredity and variability - the main features of life - is due to its great influence on other sciences. The practical nature of genetics depends on its use for practical purposes, in selection, in health care. One of the basic concepts of modern genetics is a gene. A gene is a unit of storage, tracking, and execution of genetic information. After 1900, the mechanisms of heredity began to be studied, resulting in the discovery of genetic code, transcription, translation, and the mechanisms of function of proteins encoded by specific genes. At present, the study of the delicate structure of genes in genetics "is the study of complex problems such as the management of gene activity in the individual development of the individual; until the second half of the XIX century, the phenomenon of heredity was not studied at all. But at that time it was known that children do not always look like their parents, that certain diseases occur in certain families, and that the properties of plants and animals can be improved by

mixing, some empirical laws in the field of medicine were cited. . For example, hemophilia was found to affect not only children but also mothers (Nasse's law). But the development of human genetics has always been influenced by social and political relations. Even today, it was difficult for anthropogenetics to remain a "pure science." The first notions of heredity can be found in the works of ancient Greek philosophers of antiquity. For example, Hippocrates wrote: "The seed forms the whole body, the healthy parts of the healthy seed, and the diseased parts of the diseased seed. A bald child, a blue-eyed child, from the clay to the clay, from the egg-shaped head to the egg-headed children are born. " Both Anaxagoras and Aristotle's views on heredity and gender formation are noteworthy. In his book Politics, Plato explains how to choose couples, how to raise children, for the birth of physically and mentally healthy children.

Methods of medical genetics

The basic research methods of medical genetics, which are part of anthropogenetics, are used in a manner adapted to medical practice. Francio Galton's contribution to the development of methods for studying human genetics is great. He made extensive use of genealogy and twin methods in his research, and founded biometric genetics in collaboration with his student K. Pearson. Nowadays, the number of methods of medical genetics has increased significantly, including even the latest methods of molecular biology.

In his 1865 work, Inheritance of Talent and Character, Galton studied the biographies of the most famous people of his time and found that their great success in their chosen professions was largely due to heredity. Long before Galton, there was some information about the inheritance of human traits. Moperti (in the eighteenth century) found polydactyly to be more common in some families. Nasse (early nineteenth century) proposed an empirical law of the inheritance of hemophilia.

Humans have many shortcomings as an object of genetic testing. The most important of these is that it is not possible to use hybridization in humans, i.e. the method of experimental hybridization. Nonetheless, different types of marriages can be found in a person’s society, and should be examined consistently to find the desired marriages. It is also more difficult to gather accurate information about distant relatives, without which it is impossible to create accurate genealogical maps should be studied. The large number of human chromosomes (23 pairs) is also one of the inconveniences. However, due to the success of the Human Genome project, which is currently being conducted worldwide, human chromosomes and their groups of attachments have been much better studied. Thus, modern methods of anthropogenetics allow to collect a wide variety of comprehensive information about human genetics.

Genealogical method

It is a method based on the genealogy of generations and is the most convenient method, widely known for practicing physicians. This method is the proband (the first person to be studied under the supervision of a geneticist). It is based on the collection and analysis of information about relatives. In this case, the phenotypic manifestation of the inherited, studied trait (disease) in the proband may not be observed. Although this method seems more convenient and easy, it also has some conveniences. One of them is the distortion of the information collected. If there is a hereditary latology in the family, it is natural for every family member to feel guilty about the bull. Therefore, sometimes the person in question hides the presence of Igundai disease in his offspring, or about the presence of such a disease in the offspring of her husband (ho-tininkng) gives incorrect information. The doctor should try to relieve the proband or his relatives from guilt, to build self-confidence. Whenever possible, family members should be examined by a physician in person, not limited to information provided by relatives. It is possible for such a person. if not, data are collected through questionnaires, especially if relatives live elsewhere. It is advisable that the questionnaire be completed by family physicians, Dr. Gendrix himself can see small anomalies of various development (RKA or stigmas) during the examination, and when compiling the questionnaire, the questions should be written in such a way that even non-specialists can answer correctly. It is not possible to ask if there is hypertelorism, for example), because not all doctors are well aware of what stigma this is. About 150 species of stigmas have now been identified, and stigmas may be a marker of hereditary predisposition to disease, evidence of dysembryogenesis, low penetration or expressiveness of a mutant gene. The appendix lists the types of major stigmas (see appendix).
Only complete and reliable information will allow you to create a family tree correctly.

Commonly used symbols are used to draw a tree diagram.
1 Using the genealogy method allows you to answer the following questions.
1. Whether the trait (disease) being studied is hereditary or not.
2. What is the type of heredity.
3. Forecast of the next generation.
4. In addition, this method can be used to study the intensity of mutational processes in human populations, to conduct medical-genetic counseling, to select and map mapping and interaction of genes.
The method of genealogy is carried out in 3 stages:
1) data collection;
2) creation of a family tree;
3) genealogy analysis.
Creating a tree starts with a proband. Then his relatives of I, II, III, IV levels asked and checked, non-blood relatives are also checked. In medicine, this method is also called the clinical-genealogical method, because the tree is formed in relation to any disease. For example, when studying the inheritance of hypertension, small members of the family - children - may be studied. Naturally, they still do not have

hypertension. Therefore, it is possible to test children with exercise and determine how long it takes for blood pressure to return to normal. Thus, the regulation of blood pressure is studied. The expression of the tree, that is, the description of the characters under study, must show that each member of the tree is related to the probavd.

After the data is collected, the family tree is plotted graphically, followed by one of the options for genetic analysis - genealogical analysis. It should be borne in mind that the sign (disease) occurs more than once in the family tree, but it may not be hereditary. . Or the right environmental factor may have had a detrimental effect on all members of the family. If it is determined that the trait is hereditary, proceed to determine the type of inheritance. In this case, the tree data is calculated statistically. These methods are described in detail in the monographs of K. Stern (1956), V. Makyusik. Statistical processing should begin with determining the methods by which the blood was registered (complete, incomplete, repeated). To do this, use the tables in the above manuals. Symbols are codominant (for example, AVO, MI blood groups); autosomal-dominant (e.g., brachydactyly first described in 1905), autosomal-recessive (xeroderma pigment resulting from DNA repair after exposure to ultraviolet light), or heterosoma-dominant, heterosome-recessive type.

The following characteristics can be observed in autosomal dominant inheritance:

a) the sign is observed in the majority of family members, in each generation, of the "vertical" type;

b) both parents can pass the mark on to their children in the same way;

c) the sign is the same in men and women;

g) if the sign is rare, it can be observed in about 50% of the offspring.

Characteristic features of autosomal recessive inheritance type:

a) the number of patients in the family tree is low;

b) characters are inherited in “horizontal” tits. It may not be observed at all in some generations;

c) sick children can be born from 25% of phenotypically healthy parents;

g) healthy children can be born if one of the parents is ill;

d) The sign may be the same in men and women.

Analysis of such genealogies reveals that marriages between blood relatives are more common in families where the trait is phenotypic than in the general population. It should be noted that in the presence of an epistatic gene, it can change.

Attached to the X chromosome, the specificity of heredity is that the male organism is hemizygous to the X chromosome genes (because it has only one X chromosome), passing the mark only to its daughters.

The following features are observed in the type of dominant inheritance attached to the X chromosome:

a) the sign is more common in women than in men;

b) if the mother is a carrier of the gene, it can be passed on to all children (when the mother is homozygous), or can be passed on to half of her children (when the mother is heterozygous).

c) If the sign is on the father, it will appear on all daughters, all sons will be healthy.
g) If the parents are healthy, all children will be healthy.
The following specific features are distinguished in the type of recessive inheritance attached to the X chromosome:
1. The disease is more common in men;
2. The son never receives the mark of the father.
3. If the proband is a sick woman, all her daughters and sons will be sick.
4. All children born out of wedlock to a sick man and a healthy woman are healthy, but boys born to their daughters may be sick.
5. From the marriage of a healthy man and a heterozygous woman, 50% of boys can be born sick and all girls can be healthy.

TWIN METHOD

This method was also proposed in 1875 by F. Galton. But Galton did not distinguish twins into single-egg (monozygotic-MZ) and double-egg (dizygot-DZ). Later, this method will be greatly improved by other scientists. The essence of the method is that first MZ and DZ twins and control groups from the general population are formed. Then the individual MZ and DZ are compared with each other, then the MZs are compared with DZs, and finally the twins are compared with the control group. This method determines the relative role of heredity and environment in the development of the character (disease). However, before analyzing the concordance (similarity) and discordant (dissimilarity) of the characters, it is necessary to determine which zygote (monozygotic or dizygotic) of the twins. MZs are independently developed children that divide from one zygote into two in the early stages of maturation. Therefore, the genotypes of MZs are completely similar to each other (excluding the possibility of somatic mutations), and the differences that occur in MZs are the result of environmental influences. DZs are children who develop from two simultaneously fertilized egg cells and are born at the same time. DZs may or may not resemble each other like normal sibs in the family because their total genes are 50%. They differ from other sibs in that they are under the influence of the same environmental factors in embryogenesis and after birth. If they are of different sexes, they can be added to DZs immediately.
To determine the zygote of same-sex twins, they should be compared according to different criteria. The most obvious method is a skin transplant. Good attachment of the transplant indicates monozygosity.
Comparison of NLA-system haplotypes in twins also gives good results in determining zygote. Since this system has a very large polymorphism, it is not possible to be a concordant of DZs on NA haplotypes. When comparing accurate data on the zygote of twins, other monogenic hereditary polymorphism traits, such as erythrocyte blood systems (AVO, MK, RH Lutheran, Duffy, Kidd Levis, etc.), serum systems (haptoblobin, transferrin, gamma globulin) However, it should be

borne in mind that the information obtained when the parents of the twins are identical on these traits is not accurate.

CYTOGENETIC METHOD

This method has been used since the 50s of the twentieth century. During these years, a method of artificial growth of leukocytes was developed, which introduced methods for obtaining the metaphase status of chromosomes and microscopic examination of chromosomes. The first successful outcome of this method was the discovery in 1959 by Leje of an anomaly in the number of chromosomes in Down syndrome. Currently, the cytogenetic method is widely used in the study of diseases associated with changes in the structure and number of chromosomes, mapping of chromosomes, the study of their polymorphism and the identification of other genetic problems. Only the cytogenetic method allows diagnosis in chromosomal pathologies, so this method is very convenient in differential diagnosis. For example, when the clinical manifestations of Down's disease are identified, only the cytogenetic method can be used to distinguish the trisomy variant (47, XX+21) from the translocation syndromes (45XX+T15+21). Determining this is very important in determining the prognosis of the offspring in a family with a sick child. The main stages of application of the cytogenetic method are:

1) cell separation and artificial growth;

2) obtaining the metaphase state of chromosomes;

3) microscopic study of chromosomes in the metaphase state and determination of karyotype.

Methods for separating chromosomes have also been developed in the interphase, but their exact structure can only be studied in mitosis or meiotic metaphases. Peripheral blood leukocytes are grown artificially in an environment with the addition of FGA (phyto-hemagglutinin) for 2-3 days. FGA accelerates cell division. During the metaphase stage, the chromosomes look very good, so we also add the substance colchitin to the nutrient medium with the cells dividing. This substance breaks down the dividing duct strands and stops mitosis in metaphase. A hypotonic solution is then added to the cells to break down the nuclear membrane fixed, the cells transferred to the cells of the object are stained and examined under a microscope. To separate the chromosomes, first a microphotograph of them is prepared, and then a karyogram (idiogram) is drawn. Classification of chromosomes based on the Paris nomenclature is used for individual differentiation of chromosomes. Chromosomes are mainly divided according to their length and the order of placement of centromerasinin (metacentric, submetacentric, acroceitric, satellite and other types).

At present, autoradiography, examination with 5 bromdeoxyuridine, differential staining with fluorochromes (Q), Gimza dye (S; - discs), the use of various modified variants of these methods to chromosomes in certain groups (A , V, S, D, Ye, F, J).

PALMASCOPY studies the pattern of patterns on the skin of the palm. The external structure of the cat and the patterns and images on the skin of the palm are much more complex. The palm has many cushions, folds, pads, and round stripes. There are usually 11 pads on the palm, which are divided into 3 groups: 1) 2 large proximal palm pads - tenor and hypolenor; 2) 4 finger pads; 3) Pads on 5 fingertips. From the folds in the palm: 1) palm-finger; 2) distal; 3) proximal; 4) fold of the thumb; 5) the wrist is located in a twist (Fig. 78, a). In some cases, the proximal and distal folds are combined to form four finger folds. The formation of such a fold is hereditary and often occurs in people with a different number of chromosomes (on chromosomes 13, 18, 21 and in the case of "cat scream"). There are 14 more sites in the palm. The 1st platform is located in the tenor, between the 1st and 2nd fingers between the 13th platform, the 2nd platform between the tenor and the hypotenor, the 3rd platform in the hypotenor, and the 4th and 5th platforms are located on the front of the palm. The 6th, 8th, 10th, and 12th grains are located at the base of the 5th, 4th, 3rd, and 2nd fingers, and are called the fingertips. Pads 7, 9, and 10 correspond to the space between the fingers 4, 3, and 2. The point where the three-way lines intersect is called a triradius. The palm has 4 triradiuses (a, b, c, d) at the base of the 2-4 fingers (Fig. 78, d). There is also a primary (arrow) triradius between the tenor and hypotenor. This triradius can also be located in the corpus, medial, and central parts of the palm. To determine where the main triradius is located, it is necessary to determine the angle (<atd) between the finger triradiuses and the main triradius t with d. If <atd is less than 40 °, the location of the triradius is corpuscular (t), 41-60 "is intermediate (t '), and if 6l ° and greater is central (t"). In people with inherited diseases, the primary triradius is usually located on the distal side of the palm, i.e., in the form of intermediate or central triradiuses.

DACTILOSCOPY examines the image of lines on the skin of the fingertip. The image of the lines on the fingertips is also varied (Fig. 79). They can be divided into three types: I) arched - (A - arch); 2) loop (L - loop); 3) circular or nodular (W - who). Bow-shaped images are less common than others (6%). Loop images are the most common (60%) and occur in two different directions, i.e., radial and ulnar. In the radial loop image, the lines that form it open on the radial side of the palm, while in the wrist they open on the ulnar side of the palm. Circular images are less common than loop images (34%).

Image of lines on the skin of the finger:

1 - arched; 2 - looped; 3 - transition from ilnwqsimon to circle; 4 - double loop; 5 - circular.

The ridges forming the triradius are shown to be thicker.

The triradius is not colored in the arc image. One trtradius is painted on the loop and two on the circle. The image of clips on the fingertips changes in people with inherited diseases. Once the above image is detected on the skin of the fingertips, the number of lines that protrude from the skin in them is determined. For the buniag, a line is drawn from the finger triradius to the center of the image at the tip of the finger. This connecting line is considered to have crossed several of the bulging lines on the fingertip. Since there is no triradius in the arc-shaped image,

the number of lines in it is assumed to be 0. In the loop image, the number of lines reaches a maximum of 25. Since the triradius is two in a circular image, the lines on each Lkkala side are counted, and the one with the largest number of lines is taken into account. All, that is, the lines on the fingers of the Father are 130-150 in males and 110-135 in females. Typically, when the number of X chromosomes decreases, the number of strands increases, i.e., XO-178; As the number of X chromosomes increases (e.g., XXXY), the number of strands decreases - 43. The production of images on the skin of the fingers is controlled by special genes. The circular image is said to be formed by the genes on the chromosome belonging to group E (17-18), the loop image (L), the genes on the chromosome belonging to group G (21), and the circular image on the genes on the chromosome D {13, I5). On the X chromosome, a modifier of these genes is located, which allows for a more curved image. The appearance of patterns on the skin appears at 10-19 weeks of ernbional development. By 20 weeks the erabrio skin will have all the streaks. All images on the skin are fully formed by the 6th month of the embryo and remain there for the rest of its life. When tcri is destroyed (burned, cut, etc.), the lines in them are restored as before. Today, dermatoglyphics is widely used in the detection of hereditary diseases, forensic medical examination to determine the identity, mono or dizygoticity of eguacs and ethnic composition of the population.

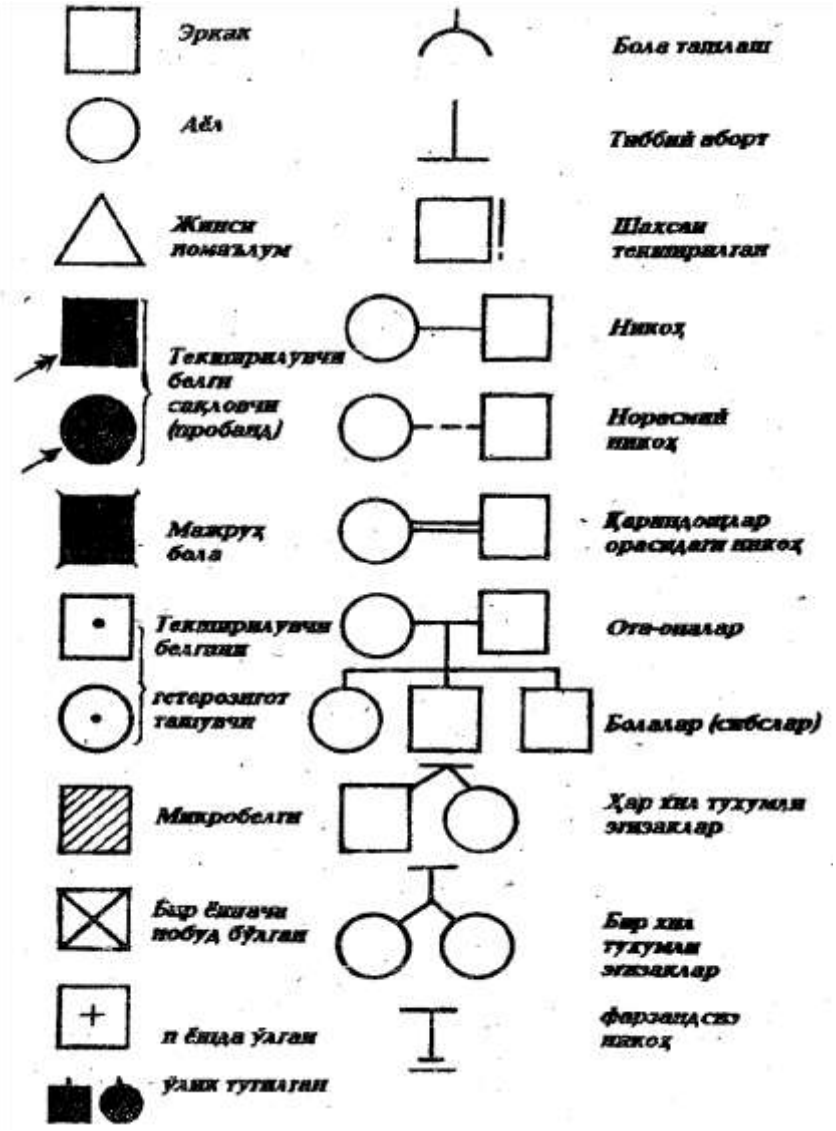
Appearance of burcliac in normal and inherited disease:

1 - Palau syndrome; 2 - Down syndrome; 3 - Shershevskiy-Temer syndrome; 4 - norm; 5 –

Kleinfelter syndrome.

The most common dermatoglyphic symptoms in people with inherited diseases are: 1) an increase in the incidence of one of the images on the fingertips (curved, looped, circular); 2) the number of protruding lines on the skin at the ends of the barmotj exceeds or decreases above the norm; 3) the main iriradiusninj in the palm: the position changes (A.) there is no triradius at the fingertip 5) a four-fingered transverse flexible fold appears in the palm.

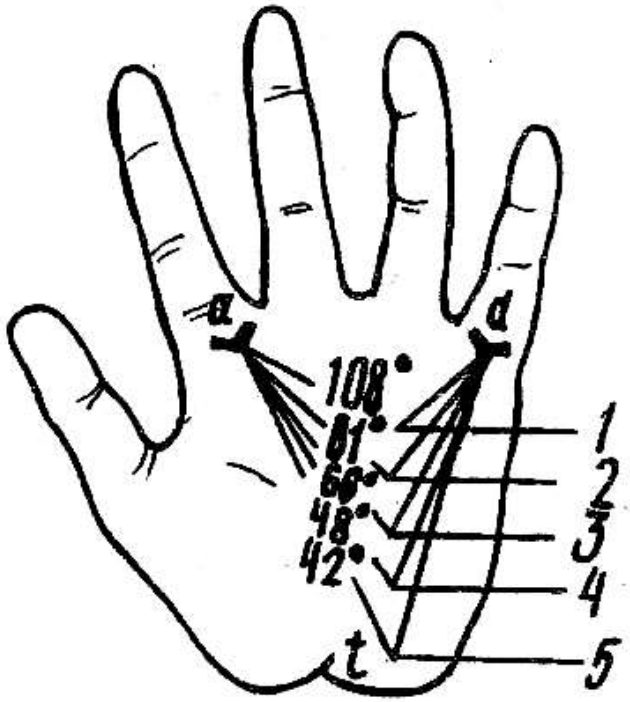
Characters used to create a family tree:



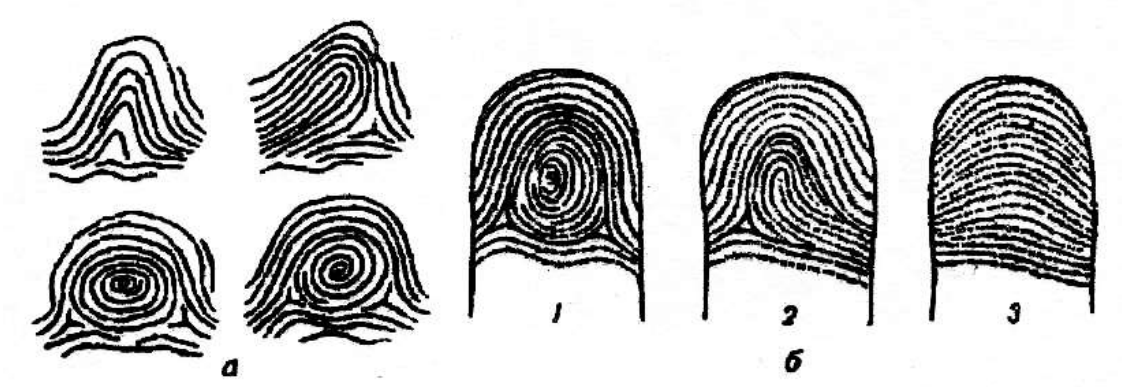
Changes in the angle of atd triradiuses in normal and chromosomal abnormalities.

1-Patau syndrome, 2-Down syndrome, Z-Shereshevsky-Turner syndrome),

In the 4th norm, in the 5th Kleinfelter syndrome.



Schematic representation of the dactyloscopy method.
schematic representation of the types of a-fingerprints, b-papillary lines.
1-circular (Urasimon) 2-circular, 3-arched



Hereditary and congenital pathologies play a major role in the general diseases of the population

According to the WHO, 5% of newborns have a genetic disorder. 40% of hereditary factors lead to death and young disabilities in early infancy. Congenital malformations account for 19% of perinatal deaths. This is due to the unsatisfactory structure of the environment and congenital hereditary diseases. The etiology of all inherited diseases is based on mutation. The process of mutation is induced and spontaneous. Damage to somatic cells is called a somatic mutation, and damage to germ cells is called a generative mutation. Mutations are divided according to the damage to the hereditary apparatus: gene, genome, chromosome. Specific hereditary diseases are divided into chromosomes and monogens. The

incidence of chromosomal diseases is 0.6-1. the risk of each child developing chromosomal pathology does not exceed 1%. Sexual and mental development is slow in autosomal disease, often ending in death in childhood. Sexual chromosomal diseases are not life-threatening and cannot always be diagnosed at birth. Medical genetics uses biochemical, clinical genealogical, dermatoglyphic and other methods of diagnosis of cytogenetic twins. In the treatment of chromosomal diseases, mainly symptomatic treatment is used.

Chromosome plays an important role in the prevention of diseases: improving the etiological conditions, hereditary status, especially the condition of women of childbearing age, the fight against harmful habits.

Diseases of chromosomes depend on changes in their number and structure. When the number changes - the genomes, when the quality changes - it is called a chromosome mutation.

According to the latest data, 0.7% of live-born babies are born with chromosomal disorders. The number of mutated embryos is even higher, however, they die during embryonic or fetal development. 40% of fetuses with spontaneous abortions and 6% of stillbirths are born with chromosomal disorders.

If we calculate the number of diseases with developmental defect defects as 100%, chromosomal diseases are detected in 35-40% of them.

Phenotypic manifestations of chromosomal diseases are caused by many defects that occur due to developmental disorders in the embryonic period. Button defects can damage many systems. As a result, pathological changes often lead to death or chromosomal disorders.

Changes in the number of chromosomes are caused by their disruption in hematogenesis in stages 1 and 2 of meiosis in daughter cells, or in the first stage of disruption of the fertilized egg cell.

Chromosomal diseases that are passed from generation to generation are mainly caused by mutations in the genome or chromosome in the somatic cells of the parents, which are passed on to the mature gametes. The pathomorphological characteristics of the disease are of great importance in the diagnosis of chromosomal diseases. However, even in this case, the main diagnosis is the cytogenetic examination.

In the treatment of chromosomal diseases, mainly symptomatic therapies are used. Symptomatic treatment can be conservative and surgical. In general, patients are recommended drugs that significantly facilitate their survival, surgical correction of button defects, physiotherapeutic methods.

What the phenotype of an organism is, the degree of development of its traits, its genotype, and the origin of genetic information do not depend on environmental conditions, but on the type of modification that occurs in a phenotype as a result of changes in external conditions. This type of variability is manifested in organisms with the same genotype as a result of their living in different conditions.

The limit of modification variability depends on the genotype, genetic characteristics of the organism, the norm of the reaction of the body's ability to change under the influence of environmental factors to a certain extent depending

on the genotype. Characters in the body vary with the degree of susceptibility to modification variability.

An example of a modifying variability is the color of the fur of protective rabbits. Rabbits of this breed have red eyes because there is no pigment in their eyes, and their bodies are covered with white hair, but the hair on their legs, ears, and tail is black because they do not have pigment. These symptoms occur only under certain environmental conditions. Hetopia of rabbits can produce pigment only at true temperatures. Therefore, in which part of the rabbit's body the blood supply is reduced, a black pigment appears. If we remove the wool from a certain part of the white rabbit's fur and carry the growth of wool from there at a low temperature, black wool will appear instead of white. If the black hairs of the rabbits are shaved off and tied in a warm place, the newly formed hairs will be white again.

If monozygotic twins with the same genotype are exposed to different climatic conditions, they may also have different changes. This means that the occurrence of traits depends not only on the genotype, but also on the environmental conditions in which the genotype is present. The color of the human eye depends on whether one is born a boy or a girl, and of course on the genotype, but the amount of pigment formed in the skin under the influence of sunlight depends on the environmental conditions.

The occurrence of a trait depends on the genotype's susceptibility to a particular environmental factor. Therefore, not all people in a given area become ill with infectious diseases. The disease occurs only in people with a predisposition to the disease in the genotype.

The variation in weight observed in animals and humans, as well as the length of the neck, is a modification variable.

Phenocopy (similarity-phenonization) can also be obtained for modification variability. Phenocopy is the process by which a change in a particular genotype under the influence of the external environment is similar to a change in another genotype. Phenocopy occurs due to physical, chemical, and biological effects. Some infectious diseases in the mother (measles, toxoplasmosis) lead to a change in the child that is similar to various inherited diseases. Because the mother's body is the environment in which this embryo grows. The presence of phenocopy makes it difficult to diagnose certain diseases.

For example, a baby's opacity may be the result of a recessive inherited disease, measles, or the mother receiving radiation during pregnancy. Deafness (deafness) caused by measles is similar to hereditary deafness.

By expressing the change in the observed signs in numbers, it is possible to get a complete idea of their variability. To do this, the studied (numerically) character index is placed in sequence from the smallest to the largest, which is called a variational series.

Typically, the variables studied in terms of numbers are divided into two: continuous, continuous. In a continuous variable, each number obtained differs very little from the other. If they are arranged in a row starting from the smallest, a continuous row is formed. Therefore, this variable is called

continuous. This includes all measurable characters. For example, a person's weight, height, etc.

In the case of a continuous variable, the magnitude of the variable placed in the variational row is now represented by a number. Therefore, this variable is called intermittent or discrete. This includes all characters that can only be represented and counted by whole numbers.

In both variables, the beginning of the character calculation is the same, and the smallest and largest value of the variable eg, i.e. the scale of the variable, is determined.

LECTURE №2:
Monogenic diseases. Criteria, hereditary type of character. Criteria and nature of multifactorial diseases.

1.1. A model of educational technology.

Class time - 2 hours	Number of students: 18
Class type	Lecture
Plan	Monogenic diseases, criteria, character, hereditary type. Criteria and nature of multifactorial diseases. FKU, alkaptonuria, galactosemia etiopathogenesis, clinic, diagnosis and treatment methods. Medical genetics advice. Methods of prenatal diagnosis. Menden's hereditary diseases.
The purpose of the topic: - During the clinical practice, the patient should be able to personally examine the patient, talk to him and provide communicative assistance. During this period, the student must live the life and mood of the clinic. Examines the patient and evaluates the internal and external organs on the basis of the propaedeutics of internal medicine. Assess the patient's condition and help improve his or her condition. The student must behave like a doctor in the presence of the patient.	
The task of the topic: - Diagnosis of major monogenic diseases. Conducting treatment and prevention of patients in outpatient and home settings after genetic disease	
Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2. Lecture Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment 5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They are divided into small groups They watch They participate They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Chromosomal diseases are inherited diseases and conditionally the hereditary mutational structure belongs to monogenic diseases. Their pathogenesis includes changes in the DNA chain, ie enlargement or loss of purine or pyrimidine bases, changes in their location, changes in nucleotides, and so on. Such changes do not affect the number and morphology of the chromosome.

Monogenic diseases are related to Mendel's law. In addition, they are characterized by clinical polymorphism, genetic heterogeneity, progredent. Multifactorial diseases play a major role in the etiopathogenesis of the main pathologies of humans (90%). Genetic endo and exo factors play a role. Such diseases cause limited heart defects in young children (pylorostenosis, urinary and genital defects, gastrointestinal tract defects, congenital anomalies, prenatal) MFZ can occur at any age. The incidence of proband in relatives depends on various factors (sex, age, birth rate, severity of the disease, the number of patients in the family, etc.)

The main task of the method of prenatal diagnosis is the diagnosis of hereditary diseases and their treatment in the womb, as well as the prevention of the birth of a child with severe hereditary pathology. Methods of prenatal diagnosis: chorionic biopsy, ultrasound, phytoscopy, aminosynthesis. Medical genetics advice will be:

-retrospective (postpartum counseling)

-prospective (counseling before the birth of a sick child)

The consultation on medical genetics is conducted in three stages. The methods should not be complicated and should occupy a large number of controllers. In our country, screening tests are performed for all people born with FKU and hypothyroidism.

The cause of gene disease is a change in the gene structure as a result of a mutation. Gene diseases can be divided into two groups:

1. monogenic diseases - (monofactorial, i.e. single factor) caused by a change in a single gene.

2. polygenic diseases - (multifactorial, i.e. multifactorial) caused by mutations in several genes.

Monogenic diseases are in complete agreement with Mendel's laws.

If we assume that a person has about 100,000 genes, there may be hereditary diseases that are caused by the same amount of genes. According to modern data, a gene contains at least 500 nucleotides, which can be mutated several tens or hundreds of times.

Genetic diseases are divided into 3 groups depending on the damage of proteins:

Injury of group I structural proteins

Injury of group II-transport proteins

Injury of group III enzymes

The third group, i.e. enzymopathy, is a relatively well-studied monogenic disease.

Enzymopoties are often caused by the absence or deficiency of a specific enzyme and lead to a slowing or cessation of biochemical reactions. Enzymopotia is caused by mutations in genes that enable the synthesis of a corresponding enzyme. In most cases, enzymopathies are accompanied by mental retardation.

The incidence in newborns is 0.1-0.5%. However, the incidence of hereditary diseases varies considerably in relatively different populations and in different geographic regions. These depend on a number of factors: isolation, environmental influences, and selection factors.

Gene disease in several groups differs depending on which metabolic disorder is involved. Hereditary diseases caused by disorders of carbohydrate metabolism, amino acids, lipids, circulating proteins (hemoglobinopathy, disorders of protein structure, etc.).

Disorders of amino acid metabolism. Among the hereditary defects of metabolism, hereditary aminoacidopathy is the largest group. Generational transmission is autosomal recessive. The development of the disease occurs due to a deficiency of one or another enzyme that responds to amino acid catabolism or anabolism.

Phenylketonuria This disease was first diagnosed in 1934 by Norwegian physician F.Fyoling. This is why it is often referred to as Fyoling’s disease. Biochemical examination of the urine of two children who were lagging behind in the development of phylloxera revealed the presence of a specific odor, a large amount of phenylpyrovinogradic acid. (stained green when Fe Cl3 is added to the patient's urine.). this substance is not found in a healthy person. It is now known to be an inherited disease. Hereditary deficiency has been reported to occur as a result of phenylalanine hydroxylase deficiency, which catalyzes phenylalanine ingested with food to tyrosine.

In the first months of life, the baby often has no symptoms and develops well in terms of weight. Interestingly, 90% of children have white blonde hair, white body and blue eyes (the frequency of encounters is in most European races). Some

babies are loose, sleepy, weak from birth, do not pay attention to toys, do not turn to sounds.

The first sign of the disease is vomiting. From the age of 2.5, a child's growth retardation is more pronounced than in others, and the child's feelings (emotions) decrease. They are not interested in anything, their attention is diminished, they do not aspire to parents and other children, and their physical development is also reduced. Sick children are much later than their peers in being able to sit or stand or walk. Teething in such children is delayed by 11-12 months.

In children with phenolketonuria, due to increased muscle tone, the body structure is disrupted: their arms are bent and their legs are bent. The patient's gait became difficult, and he was left to fend for himself. In this case, epilepsy is added.

The disease is passed from generation to generation in an autosomal recessive type. The treatment gives good results.

Treatment: Diet therapy. Phenylalanine is prescribed in a diet rich in reduced carbohydrates, mineral salts, vitamins.

Albinism This disease is caused by the absence of the enzyme tyrosinase, which converts an amino acid called tyrosine into a melanin pigment. In some patients, melanin is not produced at all or in such small amounts that a person's skin and hair become very white and their eyes become red (due to the lack of pigment, the blood vessels of the retina are not blocked). The inability to look at the light and the unusual color of the eyes make such people wear sunglasses. In equatorial Africa, an entire tribe of albinos has been bitten. There is also a village in America where people with complete albinism live. They live only at night and do not go out of their houses during the day.

American scientists conduct medical examinations of them. now a skin-based drug has been created for them. With this drug, a person can walk in the sun for 2 hours.

Clinically, albinism occurs in the following forms.

1. Eye - skin albinism
2. Eye albinism
3. Skin albinism

Eye skin has specific features of albinism. Many of them are passed down from generation to generation in the form of aphthae - recessive. Eye albinism is passed down from generation to generation, as a recessive trait associated with the X chromosome.

Alkaptonuria

In this disease, the transition process of phenylalanine and tyrazine in the subsequent manifestations is disrupted. Terosine, which enters the body through phenylalanine and food, is normally converted to R-hydroxyphenylpyravinogradic acid. This is converted to homogenesis by the enzyme. In disease, the gene that determines enzyme synthesis is mutated, so the enzyme is greatly reduced in the body. As a result, the tissues accumulate evidence of homogenetis in physiological fluids.

In this disease, the alkapton in the urine is oxidized in air and stained black. At a young age, the disease goes unnoticed, and as you get older, your symptoms begin to appear. The ridges on the necks are yellowish-purple, the supraspinatus and nasal ridges are darker.

Treatment: As a diet, the patient should consume less nutrients that are high in phenylalanine and scales. Large doses of vitamin C are given.

Disorders of carbohydrate metabolism.

Diseases caused by disorders of carbohydrate metabolism are diverse. A mutation in a gene involved in the synthesis of an enzyme that breaks down mono- di and polysaccharides in the body results in galactosymy, fructosynthesis, fentosuria, what disease, and other diseases.

Galactosymy

This disease disrupts carbohydrate metabolism. The disease is caused by impaired liver function and accumulation of galactose in the tissues (including blood). If left untreated, liver cirrhosis begins: other vital organs are also involved in the pathological process. Eventually, he dies prematurely due to illness.

With the onset of milk, the newborn's body turns yellow and relapses: dyspeptic changes occur, body mass decreases. At the early detection of the disease, children under 3 years of age are transferred to a non-dairy diet, ie dairy products are excluded from their diet. Such children are transferred to a dairy-free diet, and no changes in their psyche are observed. The number of gene carriers, ie heterozygotes, that lead to this disease averages 1: 70,000.

Practical training instructions and recommendations
Content of practical training topics

№ Lesson	Names of practical classes and their summary with the use of new pedagogical technologies
1	Methods of diagnosis of hereditary diseases. Clinical-genealogical, twin method, population-statistical method. Laboratory methods (cytogenetic, biochemical and immunogenetic methods). Dermatoglyphic method. "Step by step"
2	Chromosome diseases. etiopathogenesis, clinical, diagnostic methods. Autosomal chromosomal diseases (Down, Patau, Edwards syndrome). Sex chromosomal diseases (Klinefelter, Shereshevsky-Ternersyndromes). Case-method.
3	Monogenic disease. etiopathogenesis, clinical, diagnostic methods. Diseases of protein metabolism (FKU, cystinosis, tyrosinosis, histidinemia, homocystinuria, leukinosis, alkaptonuria). Carbon - disorders of water metabolism (galactosemia, fructosemia, glycogenosis). Disorders of fat

	metabolism (Teya-Sax Niman-Peak, Goshe disease, familial hypercholesterolemia). Molecular basis, diagnostic methods of genetic diseases, types of transmission from generation to generation, methods of treatment, prevention. "Round table."
4	Diseases of connective tissue metabolism Mucopolysaccharidoses, Marfan, Elers-Danlos and immature osteogenesis diseases. Cystic fibrosis, tubulopathies (phosphate-diabetes, De-Tony-Debre-Fanconic diseases). "Pen in the middle of the table."
5	Multifactorial diseases. The role of genetic and environmental factors in the origin of multifactorial diseases, diagnosis, treatment, prophylaxis, birth defects, ischemic heart disease, peptic ulcer disease, rheumatoid arthritis, diabetes, schizophrenia, cholecystitis, urinary tract stones, oncological, allergic diseases. "Find your partner."
6	Prenatal diagnosis of hereditary diseases. Medical-genetic counseling. Modern methods of prenatal diagnosis (UTT, chorionic biopsy, fetoscopy, amniocentesis). Guidelines for prenatal diagnosis, possible errors, complications, screening diagnostic methods. Guidelines for medical and genetic counseling, methods of calculating the risk of birth of a sick child, methods of identifying carriers of pathological genes, family planning. The Teaching-learning Method

Practical lesson №1

1.1. Methods of diagnosis of hereditary diseases.

A model of educational technology.

Class time - 6 hours	Number of students: 18
Class type	Practical training
Plan	Methods of diagnosis of hereditary diseases. Clinical-genealogical, twin method, population-statistical method. Laboratory methods (cytogenetic, biochemical and immunogenetic methods). Dermatoglyphic method.
The purpose of the topic:	

- Diagnostic methods of medical genetics; - Use of basic diagnostic methods; 1. Pay attention to patient complaints. 2. Data obtained during the collection of medical history. 3. Determination of epidemiological anamnesis data. Phase II. The goal:
Task of the topic - Use of basic medical genetic testing methods;

Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2. Practical Lesson Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment 5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They are divided into small groups They watch They participate They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Interactive method "Step by step"

This technology is designed to teach students to remember the topics covered, to think logically, to answer questions correctly and independently, and to self-assess, as well as to assess the knowledge of all students by the teacher in a short time.

The purpose of the technology is to teach students to think logically in the classroom, to express themselves freely, to evaluate themselves, to work individually and in groups, to respect the opinions of others, to choose from many ideas.

This technology can be organized individually, in small groups, or in groups during or part of a training session.

Steps:

Students will first be given time to prepare questions for the session.

Participants sit in a circle.

One of the participants is given a thread, and he says the prepared question (the student must know the answer to the prepared question), grabs the end of the thread and throws the thread to one of the students.

The student who picks up the kalava answers the question (the student who asked the question comments on the answer) and the question relay continues.

If the student does not answer the question, a ring is thrown from the string into his hand.

"The cobwebs fall"

Participants continue the question and answer session until they are all caught in a spider web.

When the students have finished asking the question, the student holding the sung kalava gives the kalava to the student who asked him the question and tells him his question, and the question is answered again until the kalava is completely rewound.

When assessing a student, the number of rings formed on his hand should be taken into account

Note. Students should pay close attention to each answer, evaluate its completeness or inaccuracy, and observe who is being targeted.

Subject statement

Diseases of chromosomes depend on changes in their number and structure. When the number changes - the genomes, when the quality changes - it is called a chromosome mutation.

According to the latest data, 0.7% of live-born babies are born with chromosomal disorders. The number of mutated embryos is even higher, however, they die during embryonic or fetal development. 40% of fetuses with spontaneous abortions and 6% of stillbirths are born with chromosomal disorders.

If we calculate the number of diseases with developmental defect defects as 100%, chromosomal diseases are detected in 35-40% of them.

Phenotypic manifestations of chromosomal diseases are the most common defects caused by impaired development in the embryonic period. Button defects can damage many systems. As a result, pathological changes often lead to death, or to the formation of chromosomal diseases.

Changes in the number of chromosomes are caused by disruption of them in hematogenesis in stages 1 and 2 of meiosis in daughter cells or in the first stage of disruption of the fertilized egg cell.

Chromosomal diseases that are passed on from generation to generation are mainly caused by mutations in the genome or chromosome in the somatic cells of the parents, which are passed on to the mature gametes.

The pathomorphological characteristics of the disease are of great importance in the diagnosis of chromosomal diseases. However, even in this case, the main diagnosis is the cytogenetic examination.

In the treatment of chromosomal disorders, mainly symptomatic therapies are used. Symptomatic treatment can be conservative and surgical. In general, patients are recommended drugs that significantly facilitate their survival, surgical correction of button defects, physiotherapy methods.

Monogenic diseases are caused by mutations in a single gene. Here, in the gene structure, one nucleotide in the DNA chain can be exchanged for another, and one purine or pyrimidine base can be exchanged for another.

According to the following general data, monogenic diseases occur in 1-2% of the total population. However, depending on the geographical or ethnic characteristics of the numbers, the encounter between different age groups can vary considerably.

The human genetic apparatus, or genome, has more than 100,000 genes. If only 1/20 of them, or 10,000 of them, can be mutated, it is not difficult to imagine how little we know about hereditary diseases.

To date, only about 3,000 monogenic diseases have been identified.

Criteria for monogenic diseases:

1. Obedience to Mendel's laws.
2. Clinical progression, ie the disease begins with mild symptoms and worsens day by day.
3. Genetic heterogeneity is the identification of clinical manifestations of diseases associated with different gene mutations.
4. Clinical polymorphism - is a variety of clinical manifestations of a single disease, the effect of treatment, the prognosis.

Monogenic diseases are passed from generation to generation through Mendel's laws:

1. autosomal dominant type;
2. autosomal recessive type;
3. sex X chromosome-dependent, dominant type;
4. sex X chromosome dependent, recessive type.

Multifactorial diseases (MFD).

MFD accounts for 90% of all human pathology. In the etiopathogenesis of these diseases, the mutation of many genes is a condition in which the environment is exposed to unfavorable conditions. These diseases do not break Mendel's laws. In monogenic diseases, the incidence of autosomal dominant diseases in the next

generation is estimated at 50%, and autosomal recessive diseases at 25%. Such a forecast cannot be made in the MFD. In their calculation, the forecast is made taking into account empirical data and many other factors.

There are specific criteria for MFD:

1) the family frequency is constantly higher than the population frequency;

2) prognosis for relatives of proband, directly proportional to the severity of his disease;

3) prognosis for relatives of the proband, proportional to the number of patients in the family;

4) from latent, mild to severe forms of the disease can be found in the family.

5) MFD is characterized by sexual dysmorphism;

6) kinship leads to an increase in marital diseases.

Prenatal diagnostic methods. Medical-genetic counseling.

Prenatal diagnostic methods are among the most effective methods of reducing hereditary diseases. Today, 100 chromosomes and about 3,000 monogenic diseases can be identified.

Methods: chorionic biopsy, UTT, amniocentesis, fetoscopy, etc.

Medical-genetic counseling is conducted in 4 stages. There are 2 methods of medical-genetic counseling: retrospective and prospective.

Guidelines and recommendations for the organization of practical training

In the hands-on activities, students will learn how to write a medical history, create a family tree, perform anthropometry, and analyze a metaphase plate.

Suggested topics for hands-on activities:

1. Methods of diagnosis of hereditary diseases (clinical-genealogical, twin method, population-statistical method). Laboratory methods (cytogenetic, biochemical and immunogenetic methods). Dermatoglyphic method.

2. Chromosome diseases. Etiopathogenesis, clinic, diagnostic methods. Autosomal chromosomal disorders (Down, Patau, Edwards syndromes). Sex chromosomal diseases (Klinefelter, Shereshevsky-Turner syndromes).

3. Monogenic diseases. Etiopathogenesis, clinic, diagnostic methods. Diseases of protein metabolism (FKU, cystinosis, tyrosinosis, histidinemia, homocystinuria, leukinosis, alkaptonuria). Carbon - water metabolism disorders (galactosemia, fructosemia, glycogenosis). Disorders of fat metabolism (Teya-Sax, Niman-Peak, Goshe disease, familial hypercholesterolemia). Molecular basis, diagnostic methods of genetic diseases, types of transmission from generation to generation, methods of treatment, prevention.

4. Diseases of connective tissue metabolism disorders (mucopolysaccharidoses, Marfan, Elers-Danlos and immature osteogenesis diseases. Cystic fibrosis, tubulopathies (phosphate-diabetes, De-Tony-Debre-Fanconi diseases).

5. Multifactorial diseases. The role of genetic and environmental factors in the origin of multifactorial diseases, diagnosis, treatment, prophylactic

methods, congenital defects, ischemic heart disease, peptic ulcer disease, rheumatoid arthritis, diabetes, schizophrenia, cholecystitis, urinary tract stone disease, oncological, allergic diseases.

6. Prenatal diagnosis of hereditary diseases. Modern methods of prenatal diagnosis (UTT, chorionic biopsy, fetoscopy, amniocentesis). Guidelines for prenatal diagnosis, possible errors, complications, screening diagnostic methods. Medical-genetic counseling. Guidelines for medical and genetic counseling, methods of calculating the risk of birth of a sick child, methods of identifying carriers of pathological genes, family planning.

Instructions and recommendations on the organization of practical classes are developed by professors and teachers of the department. In it, students enrich their knowledge and skills on the main topics of the lecture by solving practical problems. It is also recommended to strengthen students 'knowledge on the basis of textbooks and manuals, use handouts, increase students' knowledge through the publication of scientific articles and theses, problem solving, preparation of visual aids on topics, etc.

The form and content of the organization of independent work

It is recommended to use the following forms in the preparation of independent work of students, taking into account the characteristics of the subject:

"study of science chapters and topics in textbooks and manuals;

"mastering the part of reports on handouts;

"work with automated training and control systems;

"work on sections or topics of science in special literature;

“Study of new techniques, equipment, processes and technologies;

"in-depth study of disciplines and topics related to the student's academic and research work;

"Active and problem-based learning;

"distance learning.

Topics of recommended independent work:

1. Heredity and the environment.
2. Genes and destinies.
3. Dermatoglyphics.
4. Advances in Medical Genetics.
5. Social adaptation of patients with hereditary pathology.
6. Testicular feminization s-mi.
7. Methods for determining heterozygotes.
8. Cystic fibrosis.
9. Hereditary hydrocephalus.
10. What do you know about your genetics?
11. Lourensa-Muna-Pradera-Villi s-mi.
12. Social adaptation of patients with hereditary pathology.

13. Bruton's disease.
14. Hereditary syndromes and medical-genetic counseling

Evaluation criteria in the field of medical genetics

Assessment of students' knowledge in science Ministry of Higher and Secondary Special Education of the Republic of Uzbekistan dated August 7, 2009 “Regulations on the rating system of control and assessment of students' knowledge in medical higher education institutions” recommended by the Presidium of rectors of educational institutions and the Ministry of Higher and Secondary Special Education of the Republic of Uzbekistan dated August 25, 2010 333 - Amendments and additions to the Regulations and re-registered in the Ministry of Justice of the Republic of Uzbekistan on August 26, 2010 No. 1981-1 "On control over the knowledge of students in higher education institutions and Bukhara State Medical Institute named after Abu Ali ibn Sino in Neurology, Psychiatry, Narcology, Medical Genetics and Department of Medical Psychology is conducted on the basis of the "Regulations of the rating of control and evaluation of student knowledge."

The following types of control are provided to ensure that the level of knowledge and mastery of students meets the State Educational Standards:

№	Type of assessment	Max score	Sorting score	Coefficient
1.	Current evaluation	45	24,75	0,45
2.	SIW	5	2.75	0,05
4.	Final evaluation	50	27.5	0,5
	Total	100	55,0	1

a) For 86-100 points the student's level of knowledge should correspond to:

conclusions and decision making;
creative thinking;
to be able to observe independently;
to apply the acquired knowledge in practice;
understanding the essence;
to know, to tell;
to be imaginative

b) For 71-85 points, the student's level of knowledge must meet the following:

to be able to observe independently;
to apply the acquired knowledge in practice;
understanding the essence;

to know, to tell;
to be imaginative

c) For 55-70 points, the student's level of knowledge must meet the following:

understanding the essence;
to know, to tell;
to be imaginative

g) the student's level of knowledge can be assessed with a score of 0-54 in the following cases:

lack of clarity;
ignorance

The student's subject rating is determined as follows:

$$R_f = \frac{V \bullet O'}{100}$$

here:

V - the total workload (hours) allocated to the subject in the semester (cycle);

O' - Level of mastery of science (in points).

Criteria for evaluating independent work

The following forms of neurology are used in the organization of independent work of students, taking into account the characteristics of the subject, as well as the level and ability of each student's academic mastery:

- Independent study of certain theoretical topics with the help of textbooks
-preparation of information (abstract) on the given topic
-preparation for seminars and workshops
- Apply theoretical knowledge in practice
-create multimedia to create crossword puzzles and tests

Depending on the nature of the subject being taught, other forms of student independent work can be used.

Topics, forms, assignments of 38 hours for students of VI course of treatment and medical pedagogy on the subject of neurology for each subject allocated for independent work of the student are developed and approved by the scientific-methodical council of the department.

Monitoring and evaluation of student independent work.

The control and assessment of the student's independent work is evaluated on a 100-point scale from 0 to 5 points, and the rating book is added to the current assessment.

A student who scores less than 55% of the maximum grade point average for independent study will not be placed in the FC in the subject.

PRACTICAL LESSON 2

Chromosome diseases. etiopathogenesis, clinical, diagnostic methods. Autosomal chromosomal diseases

Technological map of practical training.

Class time - 6 hours	Number of students: 18
Class type	Practical training
Plan	Chromosome diseases. etiopathogenesis, clinical, diagnostic methods. Autosomal chromosomal diseases (Down, Patau, Edwards syndrome). Sex chromosomal diseases (Klinefelter, Shereshevsky-Ternersyndromes).
The purpose of the topic: - Which diseases belong to chromosomal diseases - Diseases associated with the number of sex chromosomes - The main symptoms of chromosomal diseases	
Task of the topic - - Formation of interest in the profession, humanity, a sense of responsibility; -Education of interest in expanding their knowledge, development of students' thinking skills, ogical thinking; - Guide students to a creative approach to the study of the topic.	
Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2. Practical Lesson Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic.	They are divided into small groups

	2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment 5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They watch They participate They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Related questions:

1. Describe the clinic, pathogenesis and treatment of chromosomal disease etiology
2. Describe the clinic, pathogenesis and treatment of Down syndrome etiology
3. Describe the etiology, pathogenesis and treatment of Patau syndrome
4. Shershevsky - to describe the clinic of etiology, pathogenesis and treatment of Turner syndrome
5. Describe the clinic, pathogenesis and treatment of the etiology of Klinefelter's syndrome
6. Describe the clinic, pathogenesis and treatment of the etiology of cat scream syndrome

"Modern pedagogical methods" used in teaching science

Interactive method: "Weak ring"

Required for the game:

1. A set of questions is printed on a separate sheet of paper
2. Paper with group list
3. Stopwatch

The game is on.

1. The game is played by the teacher and the assistant student is the accountant
2. The assistant student writes on a piece of paper the date, group number, name of the game and the names of the participants of this group.
3. The teacher asks the students a series of questions on a piece of paper.
4. The student must answer the question within 5 seconds.
5. The teacher evaluates the student's answer with the words "correct" or "incorrect", and if the answer is incorrect, he / she gives the correct answer.
6. The auxiliary student burns “Q” or “-” depending on the student’s last name.
7. The student is thus conducted in 2 stages of questions
8. After stage 2, the game is stopped and the student who gets 2 "-" is expelled from the game
9. The game with the rest of the students starts again on a new line

10. In another stage, questions are asked and the student who gets 2 "-" is expelled from the game. 1 "-" can also be taken from the 1st stage. If 1 "-" is obtained from the 2nd stage, the student will be expelled from the game.
11. The strongest participant in the game is selected in stages
12. In front of each surname on the paper, the teacher marks which student, at what stage, and which is the weakest link.
13. The game is evaluated with a maximum of 0.8 points.
The student who passed the 1st stage has 0 points
The student who came out of the 3rd stage answers got 0.2 points
The student who came out of the 4th stage answers 0.4 points
The student who came out of the 5th stage answers got 0.6 points
The strongest student gets 0.8 points.
14. The scores obtained by students are relevant to the current grade in the practical training.

Subject statement

Chromosomal disorders are inherited diseases in which the number or structure of chromosomes changes. A change in a chromosome is a type of mutation. If chromosome mutations occur in the early stages of the division of germ cells or fertilized eggs, they can be transferred to many cells in the developing organism, resulting in a number of developmental defects. Embryos with abnormal chromosome changes die before birth, and 6% of stillborn babies have chromosomal abnormalities.

Chromosomal disorders are mainly caused by two types of chromosome changes; in the first, there is a change in the number of chromosomes. Mac, children born with three sets of chromosomes do not live long. In most chromosomal disorders, additional chromosomes appear on one of the pairs of chromosomes (trisomy). Most often, trisomy occurs on chromosome 21, which leads to the development of Down's syndrome. In other cases, the number of sex chromosomes changes, for example, the number of chromosomes in a set of chromosomes increases to five, and the number of chromosomes increases to three. Chromosomal disorders affect the condition of the genitals and sexual maturation, and lead to infertility. Some chromosomal disorders are sex-linked. Mac, Shereshevsky Turner and Chrysomy syndrome are more common in women, and Klinefelter syndrome is more common in men, with chromosome abnormalities.

With the improvement of chromosome identification methods, many congenital malformations have been reported, particularly those associated with chromosomal aberrations. In this case, the anomalies are weaker and are associated only with certain organs and tissues. Mac, a mutation on chromosome 13 causes unilateral or bilateral retinoblastoma (cancer) in the sclera of children's eyes.

Often children with chromosomal disorders are born to healthy parents. The risk group includes mainly older women (after the age of 35-40), who have multiple births with Down's syndrome and chromosomal disorders.

Today, the Republican Screening Center in Tashkent and its branches in regional centers, where specialists conduct surveillance for early detection of hereditary diseases in pregnant women and newborns, take immediate measures to prevent chromosomal diseases. Family clinics and medical genetics clinics serve the same purpose. Year by year, the number of human hereditary diseases is increasing and new types of hereditary pathology are being identified. According to statistics, in 1956, more than 700 such diseases were detected, and by 1986 the number had risen to 2,000. By 1992, the number of hereditary diseases was 5,710, and today the number has exceeded 6,000. Usually, people with chromosomal disorders do not live long or leave offspring. Therefore, chromosomal disorders do not always pass from generation to generation. Only 3-5% of diseases are inherited and can be passed on from generation to generation. Caring for patients with this hereditary disease requires a lot of work, patience and money. The cost of caring for patients with Down syndrome alone is about the same as the cost of fighting the entire flu. Down syndrome (congenital dementia) is a chromosomal disorder characterized by severe mental and endocrine somatic changes. The disease was diagnosed by the English physician L. Down in 1866, and the disease came to be so named after him. Down syndrome is usually caused by an increase in the number of 21-pair autosomal chromosomes. The following symptoms should be considered as soon as the baby is born. Sick children are short, have small and round heads, short noses, crooked eyes, small ears, half-open mouths, and often protruding tongues. The tongue, skin, lips are dry and dull in the eyes, the teeth are not smooth, the hair on the head is sparse and smooth. The fingers are short and thick, and the fifth finger is very small. There is only one transverse branch on the skin of the palm. If the normal shortness of the atd angle in the palm does not exceed 570, it can be 800 or more in Down syndrome.

The musculoskeletal system is very poorly developed, so these children are not only mentally but also physically very weak. They don't have the ability to do things on their own. Some can be taught to read and write, but they can't learn to count, they can only do very simple household chores, and they don't have a well-developed brain. The pituitary gland, gonads, and secondary sexual characteristics develop very slowly.

The disease is caused by changes in the number of autosomes in both men and women. Girls are less likely to have menstrual cycles. People with Down's syndrome usually have no children. But it is also known that they have children, but half of the children are born with Down's syndrome. Because of their low immunity, children with Down's syndrome are more likely to suffer from various infectious diseases and malignant tumors, which can lead to death without overcoming the disease. Despite the fact that good medical care prolongs their life expectancy (30-33 years), immunodeficiency is the leading cause of death in patients with Down syndrome.

"Down syndrome". When this disease is mentioned, a person who is mentally and physically unhealthy in every way comes to mind. They have no future. What can we expect from people who are mentally weak and physically challenged ?! It is said that one should always be under the control of someone, and the life of a person who does not have the ability to control his life will be spent in darkness. Their future is often pessimistic. If there is no cure for this disease, where is the hope ?! But in this case, the assumption that depression and Down syndrome do not live longer than 25 years is not true. Because there were people with Down Syndrome who were able to have a career, to be able to manage their own lives. They can be taught. Their receptivity and mental development are developed through a special technique, which usually gives good results. Children with Down syndrome are kind, attentive, obedient, and resilient in teaching. Mental retardation (10) and some children (25 to 75) have been reported. Take a look at the pictures above: Here are some of the people who, despite being born with Down syndrome, have been able to control their will and control their lives. Pablo Pineda (Figure 1) is the first Down syndrome teacher in Europe.

The winner of the Golden Mask is Melinda Grow, an actress with Down syndrome. Hollywood actor Chris Bjork, who has Down syndrome, has also starred in several films. Yes, Down Syndrome can be a star.

The life expectancy of patients with Down syndrome is now extended. At present, the average life expectancy is 50 years. Most people with this syndrome are married.

March 21 is International Down Syndrome Day. The reason for taking this date for the third month and the 21st day of the year is due to chromosome 21 trisomy.

DAUN syndrome was diagnosed in 1866 by the English physician L. Down. Down's syndrome is usually caused by an increase in 21 autosomes. In these diseases, there are 47 chromosomes instead of 46. The disease is caused by changes in the number of autosomes and can affect both men and women. Sick children are short, small and round-headed, have short noses, crooked eye sockets, small ear supras, and often stick out their tongues out of their half-open mouths. The tongue, skin, and lips are visible and often irritated. The teeth are not flat. The hair on the head is sparsely smooth, the fingers are short and thick, and the 5th finger is very small. There is only one transverse branch on the skin of the palm. The skin of the fingertips is shaped like a loop, which opens mainly towards the ulnar.

The musculoskeletal system is also very poorly developed, so such children are very weak not only mentally but also physically. They don't have the ability to do things on their own. Some can be taught to read and write, but not to count. They can only do very simple household chores and their brains are not well developed. The pituitary gland, gonads, and secondary sexual characteristics are very poorly developed. Menstrual cycle disorders are observed in girls. People with Down's syndrome usually have no children. But it is also known to have children, but half of their children are born with Down's syndrome.

Because children with Down's syndrome have low immunity, they die at a young age without being able to cope with various infectious diseases. The cause of this

disease is still unknown. According to Beijing, Down's syndrome is more common in urban areas than in rural areas. As a mother gets older, her chances of having a baby with Down's syndrome increase.

DOWN's disease can also be caused by changes in the structure of the chromosome, i.e., as a result of chromosome translocation. In women, the majority of the 21 pairs of chromosomes are combined into 13-15 chromosomes, and in men into 20 chromosomes. As a result, the number of chromosomes in a karyotype is 45, but the genetic material is sufficient for 46 chromosomes. Therefore, this change is called balanced terracing. Theoretically, four different gametes can be formed from people with such teraslocations, and when they are fertilized with normal gametes, the following zygotes are formed.

If one of the parents has Down's syndrome due to translocation, the probability of having a healthy child in this family is very low, 33%. Down's syndrome is usually diagnosed by cytogenetic and dermatoglyphic methods. But the methods of questioning the disease are not yet clear. Edwards disease. In 1960, when Edwards identified the karyotype of a sick girl, he found that she had a single 18 chromosome ammunition (46 + 1) and studied the symptoms of this disease in detail. Boys born with Edward's disease do not live long and die in the first months of life. Girls can live up to 2-3 years. Babies with this condition are born at 3 months of age, but weigh very little. Symptoms include a swollen neck, elongated head, small jaws and mouth, high palate, very low ears, impaired circulatory system, vision, and kidney structure. The fingers are too short. There is a transverse fold on the palm, with curved lines on almost all fingertips. The disease affects 4,500 to 6,500 healthy children.

Patau disease. The disease was first studied by K. Patau in 1961. The disease occurs with an increase in one chromosome (46 + 1). This extra chromosome is one of 13-15 pairs of chromosomes, and it is difficult to say which pair it belongs to. Because 13, 14, 15 pairs of chromosomes are very similar. Therefore, Patau's disease is explained by an increase in one of the chromosomes belonging to group D. Children with this disease are usually born to healthy parents, and one sick child per 3,500 to 4,000 healthy children. Symptoms include weight, height, and premature birth. There are cracks on the upper lip and palate. There may be no eyes, the brain is not well developed, and the number of fingers is higher than usual. There are many changes in the kidneys, heart, intestines, spleen, uterus of girls, and testicles of boys. Of the dermatoglyphic signs, the principal triradius is 180 C. Sick babies usually die within 2-3 weeks after birth. Rarely can babies live up to 2-3 years. Diseases related to sex chromosomes. Patau's disease. The disease was first studied by K. Patau in 1961. The disease occurs with an increase in one chromosome (46 + 1). This extra chromosome is one of 13-15 pairs of chromosomes, and it is difficult to say which pair it belongs to. Because 13, 14, 15 pairs of chromosomes are very similar. Therefore, Patau's disease is explained by an increase in one of the chromosomes belonging to group D. Children with this disease are usually born to healthy parents, and one sick child per 3,500 to 4,000 healthy children. Symptoms include weight, height, and premature birth. There are cracks on the upper lip and palate. There may be no eyes, the brain is not well

developed, and the number of fingers is higher than usual. There are many changes in the kidneys, heart, intestines, spleen, uterus of girls, and testicles of boys. Of the dermatoglyphic signs, the principal triradius is 180 C. Sick babies usually die within 2-3 weeks after birth. Rarely can babies live up to 2-3 years. Diseases related to sex chromosomes.

Kleinfelter's disease. The disease, which occurs in men, was diagnosed in 1942 by K. Kleinfelter. In Kleinfelter's disease, the number of X chromosomes is excessive, ie 44 XXY. The ratio of children born with this disease to healthy children is 1: 1000, and this ratio is maintained even in adults.

The main symptoms of the disease are: neck, arms and legs are long, shoulders are narrow, pelvis is wide, muscles and seminal vesicles are not well developed, sperm are very small and spermatogenesis is not observed. In most cases, mental retardation occurs, and only in some cases can it be mentally normal. Images on the skin of the fingertips are often arched, with a total reduction in the number of edges. In addition to the XXY genotype of the disease, genotypes XXXY, XXXXY, XYY, XXYY, XXYYY can also occur and have a specific phenotype.

Shereshevsky-Turner disease. The disease was explained in 1925 by N.A. Shereshevsky and in 1938 by Turners. The disease is specific to women and occurs in a ratio of 1: 5000. Women with this disease have 45 chromosomes and 1 chromosome less. The main symptoms of the disease are: short stature, light weight, the neck is very short and twisted, ovarian and secondary sexual characteristics are not well developed, the shoulders are broad, the pelvis and legs are short. The menstrual cycle is not observed. The breasts do not develop and are replaced by fat deposits. The face looks older than its age. The main triradius of the palm is enlarged. Circular images are found at the fingertips. The X chromosome is often called trisomy. The disease is typically female-specific, has 44 XXX genotypes, and occurs in a 1: 1000 ratio. The phenotype can be very different. The ovaries are altered, mentally weak, physically retarded, with a narrowed yoke and high, which can leave a healthy offspring with a normal karyotype. In some cases, they are tall and the ovaries are not well developed, so infertility occurs early. Patterns on the skin of the palms and fingers have changed, but may be normal. The karyotype is almost the same in all, ie 44 XXX, but in some cases 44 XXXX and 44 XXXXX genotypes are also found. Patients with this hepatotype are more likely to have changes in their appearance.

Diseases associated with changes in chromosome structure.

Cat Scream Disease: The disease was studied by Djacobe in 1960. Later (1963) it was discovered that two children in the same family were born with the disease. It was found that the phenotypically healthy mother of these children had a deletion on chromosome 5 and that this broken part of the chromosome was attached to one of the 13th or 15th pair of chromosomes (translocation). As a result of this balanced translocation, there was little change in the mother. When chromosome 5, which is a defect in the mother, is passed on to children, children develop "cat scream" disease. If a child passes chromosome 13-15, which is a fragment of

chromosome 5, that is, a translocation, the child will not have the symptoms of the disease.

The incidence rate of children born with “cat scream” disease compared to healthy children is unclear, but it is known that the number of children with the disease has increased in recent years. The main symptoms of the disease are: changes in the vocal cords, the sound of cats screaming and meowing, mental and physical weakness, round face, small skull, antimongoloid type. 50% of patients have a malformation of the larynx, and 25% have a change in the structure of the heart. In the karyotype, chromosome 5 is significantly reduced due to deletion of the small shoulder.

Disruption in the long shoulder of the 18th pair of chromosomes.

This change in the chromosome was studied in 1964. In children with such a change in the chromosome, the skull is small, the nose is small, the larynx is narrowed, there is numbness, crooked legs, and no fingers. There will be big changes in the internal organs as well. Chromosome disease is caused by a change in the number of chromosomes or their structure. Changes in the number of chromosomes are usually caused by the unequal distribution of chromosomes at the poles during cell division.

Disruption in the long shoulder of the 18th pair of chromosomes.

People with chromosomal disorders die in infancy or leave no offspring. Therefore, chromosomal diseases are not always passed from generation to generation and reappear in each generation. Hereditary chromosomal disorders can be caused by changes in the number and structure of autosomes and sex chromosomes.

Chromosomal disorders include the following pathologies, which are clinically associated with developmental disorders. Genetically, an imbalance of chromosomal material is observed in normal cells of the organism. Many chromosomal diseases are sporadic and can occur in a healthy parent through a genome mutation or during zygote division, which is not hereditary, resulting in high mortality until the reproductive period.

The basis of the phenotype of chromosomal diseases is a disorder of early embryogenesis development. The three most common chromosomal disorders are Down syndrome, Klinefelter's syndrome, and Shereshevsky Turner syndrome.

All chromosomal diseases can be divided into two main groups,

1. Changes in the number of chromosomes, but the structure is preserved - genomic diseases.
2. Changes in chromosome structure - chromosomal diseases.

The change in the number of individual chromosomes is due to the unequal distribution of chromosomes in cells during the division of gametogenesis into one and two meiosis. This occurs in the following options:

A. Chromosome separation that is replicated in anaphase is disrupted. As a result, the double chromosome is transferred to a single female cell.

B. Disorders of conjugation of homologous chromosomes. This leads to a separation disorder of the same chromosome.

C. Lag chromosome separation during anaphase. This leads to chromosome loss. Proper diagnosis allows accurate calculation of genetic risk. But keep in mind that even when the risk is 0%, it is difficult to fully guarantee the birth of a healthy child. This is because the child may have a disease other than a genetic risk. The probability of having children with various anomalies in any family is 3-5% (overall population risk indicator). For example, when Duchenne myopathy has a relative risk factor of O%, a child with Ayer syndrome may be born into the family.

This is the result of a new mutation.

In rare cases, the risk can be as high as 100%. For example, the risk is 100% when both mothers have the classic form of autosomal recessive phenylketonuria, or both father and mother have color blindness.

There are two ways to calculate a risk indicator.

1. Determination using theoretical calculations based on genetic laws.

This method is used in monogenic diseases. Such a calculation is made taking into account the type of gametes formed, the heterozygotes in the population, with a clear parental genotype.

2. Empirical method - determination of risk indicators using tables.

This method is used for diseases caused by changes in the number and structure of chromosomes, gametes and their elimination, and for multifactorial diseases. 3. Both methods are used in combination. Such a calculation is used when one of the parents has a balanced translocation. Once the level of risk for a particular family is determined, it is multiplied by the overall population risk indicator (3-5%) and the risk indicator is assessed. Every effort can be made to assess the risk.

Risks of up to 5% are considered low, up to 10% are considered low, 20% are considered moderate, and more than 21% are considered high. However, it is not enough to determine the level of risk in childbirth counseling. In some inherited diseases, even a high risk indicator is not a reason not to have a child. For example, in color blindness, refractive anomalies, the general flexibility of the individual is almost non-existent.

It is not advisable to have a child in cases where the following risk level is higher than the general population risk level.

A. incurable hereditary diseases;

B. autosomal dominant or recessive heterosome dominant or recessive subletal and lethal gene diseases;

V. mental illness;

G. chromosomal diseases;

Usually, moderate genetic risk is the basis for not having a child.

Prenatal diagnosis is advisable in the following cases. According to the data, 5-10% of the population needs genetic counseling. That is why it is necessary to have one medical-genetic clinic for 1 million people.

Medical-genetic counseling can be done in two ways:

1. Perspective counseling is conducted when there is a possibility of childbirth, for example, when a husband and wife are exposed to harmful factors of the external environment, when a viral infection is observed during pregnancy.

2. Retrospective counseling - after the birth of a sick child in the family, is held to determine how the next child will be born.

Medical-genetic counseling is carried out in 4 stages:

In stage I, the diagnosis is determined. To do this, the client of a genetic doctor cooperation with the specialist-physician who sent you for consultation is required. All methods of genetic analysis are used to determine the diagnosis.

In stage 2, the level of risk of birth of a sick child is determined.

Stage 3 - The geneticist comes to a certain conclusion. The conclusion is prepared in writing.

In step 4, the geneticist will help the consultant make a clear decision by explaining the meaning of the face mask.

The exact structure of the prognosis depends on the accuracy of the diagnosis, the reliability of the genealogical data collected, the doctor's complete knowledge in the field of medical genetics.

Criteria for assessing the knowledge and skills of groups.

86 – 100 score	Students are fully aware of the etiology, clinic, diagnosis, classification of chromosomal diseases and are able to express independent opinions about them. Medical genetic counseling can differentiate between chromosomal abnormalities and chromosomal aberrations. Can perfectly differentiate between autosomal and sex chromosomal pathologies.
71 – 85 score	Students know the etiology, clinic, diagnosis and classification of chromosomal diseases. Medical genetic counseling can differentiate between chromosomal abnormalities and chromosomal aberrations. Can differentiate between autosomal and sex chromosomal pathologies.
55 – 70 score	Intermediate answers to questions on the topic, do not know the etiology, clinic, diagnosis, classification of chromosomal diseases. Cannot differentiate them from each other
0 – 55 score	Has a shallow knowledge of the etiology, clinic, diagnosis, classification of chromosomal diseases. Cannot differentiate them from each other.

PRACTICAL LESSON 3

Monogenic diseases. Etiopathogenesis, clinical, diagnostic methods.

Technological map of practical training.

Class time - 2 hours	Number of students: 18
Class type	Practical training
Plan	Monogenic disease. etiopathogenesis, clinical, diagnostic methods. Diseases of protein metabolism (FKU, cystinosis, tyrosinosis, histidinemia, homocystinuria, leukinosis, alkaptonuria). Carbon - water metabolism disorders (galactosemia, fructosemia, glycogenosis). Disorders of fat metabolism (Teya-Sax Niman-Peak, Goshe disease, familial hypercholesterolemia). Molecular basis of genetic diseases, diagnostic methods, types of transmission from generation to generation, treatment, prophylaxis

The purpose of the topic:

- Know how to develop risk factors,

- timely diagnosis,

- Methods of testing and identification of risk factors in young families

- To teach to use easy and convenient methods of detection of infertility in couples.

Task of the topic -

- Formation of interest in the profession, humanity, a sense of responsibility;

-Education of interest in expanding their knowledge, development of students' thinking skills, logical thinking;

- Guide students to a creative approach to the study of the topic.

Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2.Practical Lesson Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment	They are divided into small groups They watch They participate

	5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Questions on the topic:

1. Describe the clinic, pathogenesis and treatment of monogenic disease etiology
2. Describe the etiology, pathogenesis and treatment of amino acid and protein metabolism disorders
3. Describe the etiology, pathogenesis and treatment of agammaglbulinemia
4. Describe the clinic, pathogenesis and treatment of albinism etiology
5. Describe the clinic of etiology of arachnodactyly, pathogenesis and treatment
6. Describe the clinic of etiology, pathogenesis and treatment of phenyl ketanoia
7. Describe the clinic, pathogenesis and treatment of alkaptanorrhea etiology
8. Describe the etiology, pathogenesis and treatment of Duchenne muscular dystrophy
9. Describe the clinic, pathogenesis and treatment of galactasemia etiology
10. Describe the etiology, pathogenesis and treatment of essential fructosuria
11. Describe the clinic, pathogenesis and treatment of mucopolysaccharidosis etiology
12. Describe the clinic, pathogenesis and treatment of the etiology of cystic fibrosis
13. Describe the clinic, pathogenesis and treatment of sphincter myelin etiology
14. Describe the clinic, pathogenesis and treatment of mucolypidosis etiology
15. Describe the clinic, pathogenesis and treatment of hemophilia etiology

Interactive methods on the topic:

Interactive method: "Round table"

Needed for the game.

A set of questions and a situational question are presented on a separate paper

It should be individual for each student and group

Clean paper and pen

The game is on.

The group is divided into 3 small groups (4 students in each small group). Each small group sits at a separate table. He makes a blank piece of paper and takes a pen.

The date, group number, name of the game and the name of the group will be written on the paper.

One of the participants takes a question or situational question from the envelope (a question or situational question for the group is chosen by the teacher)

A separate question or situational question is selected for each group, but their complexity should be the same

A list will be distributed to everyone in the circle.

Each student writes their answer on this list.

Students will be given 3 minutes to answer each question

Over time, students hand over their work to teachers

All results are analyzed, the most correct answer is selected and the maximum score is given.

The analysis takes 15 min

All participants will receive the following points. Maximum score 0.8 points

0.8-0.7 - “5”

0.6-0.4 “4”

0.4-0.1 “3”

0- “2”

Students' scores affect the current grade in the practical class

The match will be marked at the bottom of the journal and signed by the team leader.

Student work is maintained by the teacher.

Subject statement

The classification of genetic diseases is based on three different principles: genetic, clinical, pathogenetic.

According to the genetic classification, gene diseases are autosomal-dominant (A-D),

autosomal recessive (A-R), X-linked dominant (X-D), X-linked recessive (X-R), Y-linked (golandric) and mitochondrial (cytoplasmic) diseases separated.

Clinical classification is based on which system or organ the pathological process is most often observed

depending on. For example, nerves, nerves, muscles, skin, eyes, ears, nose, throat, movement

diseases of the basic, mental, urogenital, digestive, respiratory systems. Such a classification

conditional because pathology is observed in different systems in the same disease

possible. For example, in cystic fibrosis, both in the digestive system and in the lungs

changes are observed.

Pathogenetic classification is based on the main pathogenetic chains. For example,

carbohydrates, amino acids, vitamins, lipids, depending on metabolic disorders,

disorders and diseases of metal metabolism are distinguished.

6.3. Pathogenesis of gene diseases.

The chain of pathogenesis can be described as follows: mutagenic → primary
pathological product (depending on quantity or quality) → chain of biochemical reactions
→ cell → tissue → organ → system → organism. Pathogenesis of the disease begins at the molecular level.

As a result of mutation, this gene-encoding product (protein) is redundant can be synthesized and accumulated. The second type of mutation result Anomalous protein is synthesized in the variant, which is a function of cells, organs leads to a violation. An example of this is sickle cell anemia can be cited. In the GUA triplet (codon) with the adenine of uracil glutamine amino acid instead of valine amino acid as a result of metabolism is synthesized, which reduces the solubility of hemoglobin and polymerizes it increases, reduces the binding of oxygen, causes crystallization.

Erythrocytes become sickle-shaped, agglutination increases, in the capillaries thrombi are formed, pathology of the organs begins.

In the third variant of mutant gene activity, the primary product is formed will be observed. For example, phenylalanine hydroxidase in phenylketonuria tyrosine of the amino acid phenylalanine as a result of enzyme synthesis the conversion reaction to the amino acid does not take place. Toxic effect products are collected.

The primary product in the fourth variant of mutant gene function synthesized in smaller quantities than normal. The result is a normal process slows down. Mutant allele activity disrupts the process of morphogenesis and is congenital developmental anomalies may occur (polydactyly, Meckel, Nupen syndromes, etc.) on the basis of which the process of cell differentiation is disrupted lies.

Cell-level disorders are also major in the pathogenesis of hereditary diseases important. One of the cell organelles is the activity of enzymes in the lysosome
disorders cause diseases such as mucopolysaccharidosis, glycogenosis (accumulation diseases).

Peroxisome as a result of impaired activity of peroxisome organoid enzymes diseases (osteochondrodysplasia, acatalasia, etc.)

Disorders of cell membranes in the pathogenesis of genetic diseases as well important. For example, low density in plasmolemma
Lack of lipoprotein binding receptors leads to hypercholesterolemia,
lack of androgen receptors in women despite being an XY genotype
causes the development of phenotype (testicular feminization syndrome).

Gene mutations can cause changes in cell function.

For example, oncogenic mutations can lead to colon cancer, retinoblastoma diseases develop.

data on honey were obtained by E. Elers in 1901 and H.A. Danlo in 1908

referred to as Elers-Danlo syndrome as described by.

Etiology. The disease causes the connective tissue to stretch caused by a mutation in a gene responsible for collagen synthesis (46-picture).

The main symptoms.

Excessive stretching of the skin on the skin (mainly the cheekbones, the outside of the sternum on the cheeks, wrists, knees), thinning, softening, hemorrhage, dark brown spots, numerous keloid scars, reminiscent of thin papyrus paper scars, stretch marks on the waist, legs, clear vision of veins, surgery changes such as tearing of the sutures after the treatment are detected.

Silence in the joints 90
0
more than bending in type 1 EDS, thumb to wrist straightening in type 2 EDS, elbow joint 10
0
more than bending in type 3 EDS, knee joint 10
0
bending more than in type 4 EDS, palm on the ground without bending the knee

In type 5 EDS, the fingers, wrists, and ankles are affected.

excessive bending, spontaneous protrusion of joints, like monkeys observable changes.

Changes **in the eye:** ptosis, remnants of the epicenter, retinal detachment, rupture of the eyelids.

In the ears - very elongated, flared ears.

In the teeth - partial adontia, periodontitis, multiple caries.

In the chest - scoliosis, kyphosis, lordosis, deformities of the chest (church, etiquette stone).

In the abdominal area - hernias (umbilical, white line, diaphragmatic, chov), bowel sudden perforation.

In the arms and legs - the presence of mobile nodes under the skin, blood vessels varicose expansion.

In the heart - mitral valve prolapse, arrhythmias, vegetative-vascular dystonia.

In the internal organs - gastric, renal, uterine ptosis.

What the phenotype of an organism is, the degree of development of its traits, its genotype, and the origin of genetic information do not depend on environmental conditions, but on the type of modification that occurs in a phenotype as a result of changes in external conditions. This type of variability is manifested in organisms with the same genotype as a result of their living in different conditions.

The limit of modification variability depends on the genotype, genetic characteristics of the organism, the norm of the reaction of the body's ability to change under the influence of environmental factors to a certain extent depending on the genotype. Characters in the body vary with the degree of susceptibility to modification variability.

An example of a modifying variability is the color of the fur of protective rabbits. Rabbits of this breed have red eyes because they do not have pigment in their eyes, and their bodies are covered with white hair, but the hair on their legs, ears, and tail is black because they do not have pigment. These symptoms occur only under certain environmental conditions. Hetopia of rabbits can produce pigment only at true temperatures. Therefore, in which part of the rabbit's body the blood supply is reduced, a black pigment appears. If we remove the wool from a certain part of the white rabbit's tail and carry the growth of the wool from this place at a low temperature, black wool will appear instead of white. If the black hairs of the rabbits are scraped off and tied in a warm place, the newly formed hairs will be white again.

If monozygotic twins with the same genotype are exposed to different climatic conditions, they may also have different changes. This means that the occurrence of traits depends not only on the genotype, but also on the environmental conditions in which the genotype is present. The color of the human eye depends on whether one is born a boy or a girl, and of course on the genotype, but the amount of pigment formation in the skin under the influence of sunlight depends on environmental conditions.

The occurrence of a trait depends on the genotype's susceptibility to a particular environmental factor. Therefore, not all people in a given area become ill with infectious diseases. The disease occurs only in people with a predisposition to the disease in the genotype.

The variation in weight observed in animals and humans, as well as the length of the neck, is a modification variable.

Phenocopy (appearance-similarity-phenonization) can also be obtained for modification variability. Phenocopy is the process by which a change that is related to a particular genotype and occurs under the influence of the external environment is similar to a change that occurs under another genotype. Phenocopy occurs due to physical, chemical, and biological effects. Some infectious diseases in the mother (measles, toxoplasmosis) lead to a change in the child that is similar to various inherited diseases. Because the mother's body is the environment in which this embryo grows. The presence of phenocopy makes it difficult to diagnose certain diseases.

For example, a baby's opacity may be the result of a recessive inherited disease, measles, or the mother receiving radiation during pregnancy. Deafness (deafness) caused by measles is similar to hereditary deafness.

By expressing the change in the observed signs in numbers, it is possible to get a complete idea of their variability. To do this, the studied (numerically)

character index is placed in sequence from the smallest to the largest, which is called a variational series.
In general, the variables studied in terms of numbers are divided into two: continuous, continuous. In continuous variability, each number obtained differs very little from each other. If they are arranged in a row starting from the smallest, a continuous row is formed. Therefore, this variable is called continuous. This includes all measurable characters. For example, a person's weight, height, etc.
In the case of a continuous variable, the magnitude of the variable placed in the variational row is now represented by a number. Therefore, this variable is called intermittent or discrete. This includes all characters that can only be represented and counted by whole numbers.
In both variables, the beginning of the character calculation is the same, and the smallest and largest value of the variable eg, i.e. the scale of the variable, is determined.

Changes in genetic material -mutations

The discovery of genotypic variability was the impetus for the rapid development of genetics. In 1898, the Russian scientist SI Korzhinsky discovered that there would be genetic changes, and two years later the Dutch botanist G. De Friese introduced the concept of mutation into science. G. De Friese has studied mutations in plants for many years.
In the case of a continuous variable, the magnitude of the variable placed in the variational row is now represented by a number. Therefore, this variable is called intermittent or discrete. This includes all characters that can only be represented and counted by whole numbers.
In both variables, the beginning of the character calculation is the same, and the smallest and largest value of the variable eg, i.e. the scale of the variable, is determined.
It has been tracking a plant called enotera (a plant belonging to the Cypriot family. These include the Cypress or Ivan tea plant) for 17 years. He observes many plants and first finds three mutants: one is stunted, the other is a giant with long stems, the flowers, fruits and leaves are large. The third had red veins on the leaves and fruits. The scientist developed his theory of mutation in 1901-1903 by observing and summarizing mutants for a long time. The rocks put forward in this mutation theory are:
1. Mutations occur suddenly;
2. New characters formed as a result of mutation are stable;
3. Mutations are hereditary, as opposed to non-hereditary variability. It is passed down from generation to generation;
4. The intermediate form of the variable does not form a continuous series around it. Because mutation results in a qualitative change;
5. Mutations occur in various forms and can be beneficial or harmful;

6. The probability of occurrence of mutations depends on the number of organisms studied;
7. Similar mutations can occur more than once.
The doctrine of mutation was then comprehensively developed and many types of mutations were identified. Mutation refers to the occurrence of chemical changes in genes that retain normal traits that are passed down from generation to generation under the influence of various environmental factors. Mutations can occur under the influence of external and internal environment.
Mutations involve various features of the structure and function of an organism. For example, in Drosophila, the shape of the rows, body color, the development of body hair, eye color (yellow, white, dark red), as well as many physiological characteristics (life expectancy, fertility, various harmful effects) resistance, etc.) mutation changes are known.
Mutations are constant in nature. This is a simple phenomenon of nature. Mutations often occur spontaneously and undirected, i.e., they are beneficial, indifferent, but often harmful or even lethal.
Beneficial mutations are preserved as a result of evolution, increase the viability of the species and lead to the formation of new species. Harmful ones reduce the viability of the species, leading to the formation of disabled organisms and the regression of the species. It often becomes the main cause of hereditary diseases.
Thus, as a result of the mutated gene appearing in the body of the next generation, hereditary diseases begin in a family that has not yet had hereditary diseases and begin to spread to the offspring.
Under natural conditions, the rate at which mutations form is very low. Mutations in the transmission of human genes from generation to generation range in average from 1:10 to 1:10. This means that only one in a million cells can form a new mutation. But given the sheer number of genes, at least 10 percent of both male and female gametes will have some kind of new mutation.
Mutations that occur depending on their occurrence are passed on from generation to generation in a dominant and recessive type. If a mutation of the recessive type is formed, it can be stored in latent conditions without occurring for several generations.
This only happens when such a gene is mixed with a selective organism. If a mutation occurs in the dominant type, then the occurrence of the mutation depends on the viability and reproductive characteristics of the organism. For example: achondroplasia (or chondrodystrophy) is a common disease of the skeletal system: tubular bones consist of a tumor of the pineal gland, a change in the shape of the base of the head and nasal bones. This pathology results in abnormally short limbs with normal body and head sizes. The saddle of the forehead and the saddle of the forehead make the appearance of such a person even more strange. The reproductive traits of people with this disease for social and biological reasons are much lower than normal.

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Gene diseases

The cause of gene disease is a change in the gene structure as a result of a mutation. Gene diseases can be divided into two groups:

1. monogenic diseases - (monofactorial, i.e. one factor) caused by a change in a single gene.
2. polygenic diseases - (multifactorial, i.e. multifactorial) caused by mutations in several genes.

Monogenic diseases are in complete agreement with Mendel's laws.

If we assume that a person has about 100,000 genes, there may be hereditary diseases that are caused by the same amount of genes. According to modern data, a gene contains at least 500 nucleotides, which can be mutated several tens or hundreds of times.

Genetic diseases are divided into 3 groups depending on the damage of proteins:

Injury of group I structural proteins

Injury of group II-transport proteins

Injury of group III enzymes

The third group, i.e. enzymopathy, is a relatively well-studied monogenic disease.

Enzymopoties are often caused by the absence or deficiency of a specific enzyme and lead to a slowing or cessation of biochemical reactions. Enzymopotia is caused by mutations in genes that enable the synthesis of a corresponding enzyme. In most cases, enzymopathies are accompanied by mental retardation.

The incidence is 0.1-0.5% in newborns. However, the incidence of hereditary diseases varies considerably in relatively different populations and in different geographic regions. These depend on a number of factors: isolation, environmental influences, and selection factors.

Gene disease in several groups is differentiated depending on which metabolic disorder is involved. Hereditary diseases caused by disorders of carbohydrate metabolism, amino acids, lipids, circulating proteins (hemoglobinopathy, disorders of protein structure, etc.).

Disorders of amino acid metabolism. Among the hereditary defects of metabolism, hereditary aminoacidopathy is the largest group. Generational transmission is autosomal recessive. The development of the disease occurs due to a deficiency of one or another enzyme that responds to amino acid catabolism or anabolism.

Phenylketonuria This disease was first diagnosed in 1934 by Norwegian physician F.Fyoling. This is why it is often referred to as Fyoling’s disease. Biochemical examination of the urine of two children who were lagging

behind in the development of phylloxera revealed the presence of a specific odor, a large amount of phenylpyrovinogradic acid. (stained green when Fe Cl3 is added to the patient's urine.). this substance is not found in a healthy person. It is now known to be an inherited disease. Hereditary deficiency has been reported to occur as a result of phenylalanine hydroxylase deficiency, which catalyzes phenylalanine from food into tyrosine.

In the first months of life, the baby often has no symptoms and develops well in terms of weight. Interestingly, 90% of children have white blonde hair, a white body, and blue eyes (the frequency of encounters is in most European races). Some babies are loose, sleepy, weak from birth, do not pay attention to toys, do not turn to sounds.

The first sign of the disease is vomiting. From the age of 2.5, a child's growth retardation is more pronounced than in others, and the child's feelings (emotions) decrease. They are not interested in anything, their attention spans, they do not aspire to parents and other children, and their physical development also declines. Sick children are much later than their peers in being able to sit or stand or walk. Teething in such children is delayed by 11-12 months.

In children with phenolketonuria, due to increased muscle tone, the body structure is disrupted: their arms are bent and their legs are bent. The patient's gait became difficult, and he was left to fend for himself. In this case, epilepsy is added.

The disease is passed from generation to generation in an autosomal recessive type. The treatment gives good results.

Treatment: Diet therapy. Phenylalanine is prescribed in a diet rich in reduced carbohydrates, mineral salts, vitamins.

Albinism. This disease is caused by the absence of the enzyme tyrosinase, which converts an amino acid called tyrosine into a melanin pigment. In some patients, melanin is not produced at all or in such small amounts that a person's skin and hair become very white and their eyes become red (due to the lack of pigment, the blood vessels of the retina are not blocked). . The inability to look at the light and the unusual color of the eyes make such people wear sunglasses. In equatorial Africa, an entire tribe of albinos has been bitten. There is also a village in America where people with complete albinism live. They live only at night and do not go out of their houses during the day.

American scientists conduct medical examinations of them. now a skin-based drug has been created for them. With this drug, a person can walk in the sun for 2 hours.

Clinically, albinism occurs in the following forms.

1. Eye - skin albinism
2. Do you have eyeballs
3. Skin albinism

Eye skin has specific features of albinism. Many of them are passed down from generation to generation in the form of aphthae - recessive. Eye

albinism, as a recessive trait associated with the X chromosome, is passed down from generation to generation.

Alkaptonuria

In this disease, the transition process of phenylalanine and tyrazine in the subsequent manifestations is disrupted. Terosine, which enters the body through phenylalanine and food, is normally converted to R-hydroxyphenylpyravinogradic acid. This enzyme converts homogentesin into acid. In disease, the gene that determines enzyme synthesis is mutated, so the enzyme is greatly reduced in the body. As a result, the tissues accumulate evidence of homogenetis in physiological fluids.

In this disease, the alkapton in the urine is oxidized in air and stained black. At a young age, the disease goes unnoticed, and as you get older, your symptoms begin to appear. The ridges on the necks are yellowish-purple, the supraspinatus and nasal ridges are darker.

Treatment: As a diet, the patient should consume less nutrients that are high in phenylalanine and scales. Large doses of vitamin C are given.

Disorders of carbohydrate metabolism.

Diseases caused by disorders of carbohydrate metabolism are diverse. A mutation in a gene involved in the synthesis of an enzyme that breaks down mono- di and polysaccharides in the body results in galactosymy, fructosynthesis, fentosuria, what disease, and other diseases.

Galactosymy

This disease disrupts carbohydrate metabolism. The disease is caused by impaired liver function and accumulation of galactose in the tissues (including blood). If left untreated, liver cirrhosis begins: other vital organs are also involved in the pathological process. As a result, he became ill and died prematurely.

With the onset of milk, the newborn's body turns yellow and relapses: dyspeptic changes occur, body mass decreases. When the disease is detected early, children under 3 years of age are transferred to a non-dairy diet, ie dairy products are excluded from their diet. Such children are transferred to a dairy-free diet, and no changes in their psyche are observed. The number of gene carriers, ie heterozygotes, that lead to this disease is on average 1: 70 thousand.

Mucopolysaccharidoses

This disease is caused by a disorder of mucopolysaccharide metabolism. Mucopolysaccharides accumulate in large numbers in lysosomes because the lysosome does not have an enzyme that breaks them down. With this disease, the structure of the skeletal skull, eyes, face and internal organs changes and mental weakness is observed. They accumulate in the spleen, spleen, bone marrow, and other tissues and are excreted in sand and urine. Children with the disease can live up to 12 years.

There are currently 4 types of mucopolysaccharide disease, all of which produce the same phenotypic appearance. All types of mucoposaccharidoses are inherited through a recessive gene in the aphthosome.
Type 1 is called Gurler syndrome and was described by Gurler in 1919. There are no external signs at birth (only the body weight is higher). From the age of 2 months, the body facial structure changes. His mouth is open. Mainly lives up to 10 years. It is caused by a deficiency of the enzyme L-idurolitase and a violation of the catabolism of mucopolysaccharides.
Type 2 is called Hunter Syndrome and was described by Hunter in 1917. A child born with this disease does not develop any clinical symptoms until the age of 1-2 years. Only in some cases there is rhinitis of the external respiratory organs or macro and scaphocephaly, umbilical hernias. By the age of 2, neck growth slows, body skin thickens, and short necks and teeth erupt.
Type 3 Sanfilipo Syndrome, described by Safilipo in 1963, is a milder form of the disease than others, with children as young as 2-3 years old sometimes growing to school age. Then there are changes in the psyche of children.
Type 4 is called Marcio and Brailsford syndrome and was described in 1929 by Marcio and Brailsford. From the age of 2, bone deformities are observed when growth is slowed.

Diabetes mellitus

This disease occurs as a result of metabolic disorders in the body, mainly sugar metabolism. In diabetes, blood sugar levels rise. Sugar also begins to be excreted in the urine. The amount of sugar in the urine is an indicator of the patient's severity. Type 3 Sanfilipo Syndrome, described by Safilipo in 1963, is a milder clinker than others, with children as young as 2-3 years old and sometimes as normal as school-age children. Then there are changes in the psyche of children.
Type 4 is called Marcio and Brailsford syndrome and was described in 1929 by Marcio and Brailsford. From the age of 2, bone deformities are observed when growth is slowed.
Diabetes mellitus
What the phenotype of an organism is, the degree of development of its traits, its genotype, and the origin of genetic information do not depend on environmental conditions, but on the type of modification that occurs in a phenotype as a result of changes in external conditions. This type of variability is manifested in organisms with the same genotype as a result of their living in different conditions.
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The limit of modification variability depends on the genotype, genetic characteristics of the organism, the norm of the reaction of the organism to the ability to change under the influence of external environmental factors to a certain extent depending on the genotype. Characters in the body vary with the degree of susceptibility to modification variability.

An example of a modifying variability is the color of the fur of protective rabbits. Rabbits of this breed have red eyes because they do not have pigment in their eyes, and their bodies are covered with white hair, but the hair on their legs, ears, and tail is black because they do not have pigment. These symptoms occur only under certain environmental conditions. The getopy of rabbits can produce pigment only at true temperatures. Therefore, in which part of the rabbit's body the blood supply is reduced, a black pigment appears. If we remove the wool from a certain part of the white rabbit's tail and carry the growth of the wool from this place at a low temperature, black wool will appear instead of white. If the black hairs of the rabbits are scraped off and tied in a warm place, the newly formed hairs will be white again.

If monozygotic twins with the same genotype are exposed to different climatic conditions, they may also have different changes. This means that the development of traits depends not only on the genotype, but also on the environment in which the genotype is present. The color of the human eye depends on whether one is born a boy or a girl, etc. Of course, it depends on the genotype, but the amount of pigment formation in the skin under the influence of sunlight depends on environmental conditions.

The occurrence of a trait depends on the susceptibility of the genotype to a particular external environment. Therefore, not all people in a given area become ill with infectious diseases. The disease occurs only in people with a predisposition to the disease in the genotype.

The variation in weight observed in animals and humans, as well as the length of the neck, is a modification variable.

Phenocopy (appearance-similarity-phenonization) can also be obtained for modification variability. Phenocopy is the process by which a change in a particular genotype under the influence of the external environment is similar to a change in another genotype. Phenocopy occurs due to physical, chemical, and biological effects. Some infectious diseases in the mother (measles, toxoplasmosis) lead to a change in the child that is similar to various inherited diseases. Because the mother's body is the environment in which this embryo grows. The presence of phenocopy makes it difficult to diagnose certain diseases.

For example, a baby's opacity may be the result of a recessive inherited disease, measles, or the mother receiving radiation during pregnancy. Deafness (deafness) caused by measles is similar to hereditary deafness.

By expressing the change in the observed signs in numbers, it is possible to get a complete idea of their variability. To do this, the studied (numerically)

character index is placed in sequence from the smallest to the largest, which is called a variational series.

In general, the variables studied in terms of numbers are divided into two: continuous, continuous. In continuous variability, each number obtained differs very little from each other. If they are arranged in a row starting from the smallest, a continuous row is formed. Therefore, this variable is called continuous. This includes all measurable characters. For example, a person's weight, height, and so on.

In the case of a continuous variable, the magnitude of the variable placed in the variational row is now represented by a number. Therefore, this variable is called intermittent or discrete. This includes all characters that can only be represented and counted by whole numbers.

In both variables, the beginning of the character calculation is the same, and the smallest and largest value of the variable eg, i.e. the scale of the variable, is determined.

Criteria for assessing the knowledge and skills of groups.

86 – 100 score	The student has a thorough knowledge of monogenic diseases, their etiology, clinic, is able to think independently, as well as the principles of their classification, diagnosis and treatment. Can give genetic advice. Can better differentiate cystic fibrosis from fermentopathy.
71 – 85 score	The student knows the principles of monogenic diseases, their etiology, clinic, classification, diagnosis and treatment. Can give genetic advice. Can differentiate cystic fibrosis from fermentopathy.
55 – 70 score	Answers questions on the topic at a moderate level, does not know the principles of monogenic diseases, their etiology, clinic, classification, diagnosis and treatment. Cannot differentiate them from each other
0 – 55 score	He has a shallow knowledge of monogenic diseases, their etiology, clinic, classification, diagnosis and principles of treatment. Cannot differentiate them from each other.

PRACTICAL LESSON 4
Diseases of connective tissue metabolism disorders

Class time - 2 hours	Number of students: 18
Class type	Practical training
Plan	Disorders of connective tissue metabolism Mucopolysaccharidoses, Marfan, Elers-Danlos and immature osteogenesis diseases. Cystic fibrosis, tubulopathies (phosphate-diabetes, De-Tony-Debre-Fanconic diseases).

The purpose of the topic:

- Know how to develop risk factors,

- timely diagnosis,

- Methods of testing and identification of risk factors in young families

- To teach to use easy and convenient methods of detection of infertility in couples.

Task of the topic -

- Formation of interest in the profession, humanity, a sense of responsibility;

-Education of interest in expanding their knowledge, development of students' thinking skills, logical thinking;

- Guide students to a creative approach to the study of the topic.

Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2.Practical Lesson Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment	They are divided into small groups They watch They participate

	5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Questions on the topic:

1. What is tubulopathy?
2. What is the etiopathogenesis of tubulopathy?
3. Name the clinical signs of tubulopathy.
4. Etiology, clinic, principles of treatment of mucopolysaccharidoses.

Interactive method: FSMU game

Required for the game:

1. A set of questions printed on separate papers.
2. Pure white paper
3. Ordinal numbers corresponding to the number of students in the group
4. The course of the game.
5. Total game time - 45 minutes
6. The group is divided into 3 small groups (3-4 students for each small group).
7. Each small group sits at a separate table. He makes a blank piece of paper and takes a pen.
8. The date, group number, name of the game and the names of the participants of the group are written on the paper.
9. One of the participants takes a question or situational question from the envelope (the teacher chooses the question or situational question for the group)
10. A separate question or situational question is selected for each group, but their complexity should be the same.
11. 15 min time is given
12. A small group discusses the question and writes down their opinion.
13. The assistant should make sure that students do not move and do not participate with the neighboring small group.
14. After 15 minutes, the papers are collected
15. The work is handed over to the teacher.
16. All participants are given the following points. Maximum score 0.8 points

0.8-0.7 - “5”
0.6-0.4 “4”
0.4-0.1 “3”
0- “2”

17. At the end of the answers the assistant signs and signs.
18. The score obtained by students affects the current grade in the practical training

19. In the lower part of the magazine is marked the game, the leader of the group signs.
Students' work is kept by the teacher.

Course Outline

Rickets is a common disease among children under 2 years of age. There are also cases of late development of rickets, which coincides with the period of intensive growth of the skeleton and the body.
Rickets was known long ago, and Sorana Efeysky died in 98-138. BC and Galen (131-211 BC) cocktails are remembered. In 1960, English orthopedist F. Glisson laid the clinical and pathological basis for rickets. The disease is also known as "English disease" because of its rapid spread in the UK. The English name rickets is derived from the Old English word wricken, meaning to wear, while Glisson changed it to the Greek word rachitis (spine), so rickets emphasizes the fact that the spine is worn.
In the early twentieth century, the Russian scientist I.Shabad found that fish oil is effective in the treatment and prevention of rickets.
years American researcher Mellanby found that the initial active effect of fish oil is due to the fat-soluble vitamin.
In 1922, D. McCollum discovered and obtained vitamin D, which made it possible to study its specific effects on bones, muscles, intestines, and renal canals. Rickets is common in all countries, especially in northern countries where there is not enough sunlight. Babies born in the fall and winter months are more prone to rickets, and the disease is more severe.
According to V. Osner, in Karaganda in 1928, about 50-80% of children in the twentieth century were infected with rickets in Austria and England.
In the first half of the twentieth century, 46-68% of children in Russia were infected with rickets.
In Bulgaria, where most of the year is sunny, the incidence among children under one year of age is 20%. In recent years, rickets has risen from _____% to _____% among children.
The etiopathogenesis of rickets.
One of the main and many causes of classic rickets is the ingestion of vitamin D through food or its insufficient production in the skin.

Factors contributing to rickets.

By the mother
1. Mother's age (under 17 and over 35)
2. Toxicosis of pregnancy
3. Extragenital pathologies (metabolic diseases, pathologies of the stomach, intestines and kidneys)
4. Insufficient nutrition during pregnancy and lactation (protein deficiency, Sa, R, vit.D, V1, V2, V6 deficiencies)

5. Non-compliance with the agenda (lack of insolation, hypodynamics)
6. Complicated birth
7. Asymmetry of socio-economic conditions (including in the transition to a market economy)
By the child
1. Date of birth (children born between June and December)
2. Premature birth, morphofunctional deficiency.
3. Weight gain at birth (more than 4 kg).
4. "Sudden increase in body weight" in the first 3 months.
5. Breastfeeding, but breastfeeding with “old” milk.
6. Early artificial and mixed feeding of milk not adapted to the composition of breast milk.
7. Insufficient use of fresh air.
8. Insufficient range of motion (severe walking, lack of exercise and massage).
9. Perinatal encephalopathy III with corneal lesions.
10. Skin, liver and kidney diseases, malabsorption syndrome.
11. Frequent urticaria and intestinal diseases.
12. Taking anti-epileptic drugs (such as phenobarbital, etc.).
The metabolism of vitamin D is very complex. It is now known that vitamin D (cholecalciferol) is formed in the skin under the influence of ultraviolet light of a certain length (280-310 μm-Dorno light). In Uzbekistan, the sun's rays mainly reflect ultraviolet light, but do not fall into the range of Dorno rays.
Despite the fact that there is enough sunlight in our country, rickets is very common. A good amount of vitamin D is formed in the skin of a child, depending on the weather in conditions of insufficient sunlight (frequent fog, clouds, humid air) or living conditions reduce the intensity of vitamin Dsynthesis. Therefore, the incidence of rickets is higher in industrial areas than in rural areas, especially in autumn and winter. Vitamin D enters the gastrointestinal tract in the form of cholecalciferol (D3). This vitamin DZ is related to animal products or ergocalciferol D2 is a drug.
In the liver and kidneys through complex changes in the body This vitamin D is converted into more active metabolites, which allows Sa and R to be transported in the small intestine and Sa and R to accumulate in the bones. Multi-component regulation of R-Sa homeostasis is mainly performed by parathyroid hormone, vitamin D, and calcitonin. In the case of impaired homeostasis of Sa and R, the above-mentioned metabolic events occur in various cells (bone marrow, gastrointestinal tract, liver, kidney) in the internal and external cells of Sa.
allows for quick recovery. Disruption of the function and structure of the said organ and biochemical system leads to various hypocalcemic states. To identify the main link in the Sa metabolism disorder, we need to fall into the scheme of the general management of Sa-R homeostasis.

The lower limit of Sa in the deposit is 2; high-28 mmolG`l. Giopcalcemia rapidly activates the synthesis of parathyroid hormone, which accelerates the excretion of Sa from the bone marrow, as well as the excretion of R in the kidneys, resulting in decreased reabsorption in the renal ducts and a normal ratio of Sa: R (normally 2: 1).

The second major regulator of Sa homeostasis is vitamin D. Its homeostatic action is aimed at restoring decreased Sa in the deposit, which is slower than that of parathyroid hormone. In conditions of hypocalcemia, vitamin D acts on the bone and improves parathyroid hormone-specific bone marrow transplantation: helps to increase the number of osteoblasts, reduces bone displacement and cortical porosity. Many organ cells have vitamin D3 receptors, which provide intracellular enzymatic regulation.

Hypocalcemia caused by one reason or another is the first link in complex pathophysiological processes. Hypocalcemia activates the activity of the thyroid gland and induces parathyroid hormone hyperfunction and ensures the removal of inorganic Sa from the bones. Its location in the gastrointestinal tract and kidneys is also determined. In particular, the excretion of Sa and R salts is disrupted in the small intestine, while the excretion of phosphates and amino acids is reduced in the renal ducts. The result is rapid hypophosphatemia and hypoproteinemia, leading to the development of alkaline reserve and acidosis in the deposit. Also, the increase in acidosis is due to the formation of stratum corneum.

Hypophosphatemia leads to an acceleration of R secretion from organic tissues. Primarily, this applies to phosphatides, i.e., triphosphate in muscle tissue to the myelin sheath of the cell and nerve endings.

The predominance of the excitation process depends on the demyelination, which is then replaced by a single braking reaction. In muscle tissue, energy volume is disrupted and tone is reduced. Acidosis causes universal microcirculatory disorders. As a result, pathological reactions occur in the CNS and internal organs, especially in its structure, which serves as an additional excretory organ. Increases the porosity of the vascular wall, mucous-producing gastrointestinal tract glands and lungs, which increase the oxidative products of metabolism. In acidosis again develops dystonia of the autonomic nervous system, usually vagotonia predominates. Individual metabolic disorders lead first to functional, then to morphological changes of internal organs, especially respiratory and digestive organs. Immunological protection is reduced, along with the development of a premorbid background, which contributes to the prolongation and severity of the disease. Osteogenesis is impaired as a result of changes in Sa, R metabolism.

Clinical landscape.

The development or exacerbation of the disease occurs in late autumn and winter. Self-medication can be observed in late spring and summer.

As a result of disturbances in Sa vaR metabolism and deficiency of active metabolite of vitamin D, changes are observed in the CNS, bone tissue, many internal organs. Usually the first symptom of rickets is manifested in 1-2 months of life.
The characteristic clinical manifestations of rickets serve in its basic diagnosis. Because it is a common disease, rickets is manifested by a variety of symptoms.

Symptoms in the early stages of rickets.
Central and autonomic nervous system.
Discomfort, fear, irritability, lightheadedness, loud noise, sleep disturbances, sweating (sticky) skin, increased moisture, decreased turgor, heat transfer, hair loss in the neck area.
Muscular system, constipation, muscle hypotension.
Changes in bone
The edges of the large liqueur are slightly concave.
Laboratory indicators
The norm in the Sa deposit, the norm in the R deposit, or decreased, the alkaline phosphatase in the deposit is reduced. Metabolic acidosis.
Radiological changes
It is usually undetectable.
Rickets is a series of organ damage. the initial clinical manifestations of the disease (profuse sweating, sleep disturbances, loss of appetite, red dermographism) can be observed by the parent in the late first months, early in the second months. These appearances are due to vegetative disorders. This will soon lead to changes by the MNS: restlessness, high mobility, lightheadedness, loud noise, and so on. The child's sleep worsens and he rubs his head on the pillow, as a result of which the hair on his neck falls out, he sweats a lot, and the smell of sweat is sour. The skin begins to heat up. One of the characteristic symptoms is a decrease in muscle tone and a tendency to constipation, during which the changes by the bone are almost undetectable, only the large licked edges can be detected.
No changes are detected when the bones are x-rayed. During the biochemical examination, the amount of Sa in the deposit is normal, but the amount of R in the deposit is reduced and its excretion in the urine is increased.
The duration of the initial period can last an average of 2-3 weeks to 2-3 months, and this depends on the tendencies of the child to live. Timely diagnosis and treatment of rickets can lead to a complete cure in a few weeks. If left untreated, it can lead to bone changes. In particular, the intensively growing bones during this period are severely damaged. One of the early signs in the skeletal system in rickets is CRANIOTABES. Often the posterior part of the upper bones and the nape of the neck, rarely accompanied by softening of the other bones. Another characteristic change is that the incomplete areas of the skull are sunken. Most often craniotabes

are detected in children under 6 months of age. In addition, immunity is reduced in rickets. These changes are caused by frequent inflammatory diseases of the upper and lower respiratory tract.
At the same time there is an expansion of the boundaries of the heart due to muscle hypotension. ECG changes may include a decrease in tooth voltage, an increase in the P-Q interval, an expansion of the QRS complex, and an increase in systolic values.

Symptoms in the period when the clinical manifestations of rickets are obvious.
Central and autonomic nervous system
He is sweating profusely. Increased general weakness. Lagging behind psychomotor development. Emotional lability. Muscular system
Muscle hypotension (frequent constipation). Wrinkles play.
«Baka korin». High diaphragm.
Changes in bone
Craniotabes. Flattening of the neck bone. The head is square. enlargement of the forehead and nape of the neck "Olympic forehead", "saddle nose". Disorders of tooth decay (not in the time of decay and face). Bite disorders. chest deformity (ethical chest, "chicken breast", kyphosis, lordosis, scoliosis). Variation of lower aperture (garrison skirt). Wrinkling of long tubular bones. Ricky flat powder, rosary on the ribs. Bracelet on the wrists. Branches of pearls between the gray fingers.
Laboratory indicators
The amount of Sa and R in the deposit decreased. Increased alkaline phosphatase in the deposit. The amount of R in the urine is normal or increased.
Radiological indicators
Osteoporosis. Glass-like expansion of metaphyses.
It should be noted that muscle hypotension and electrolyte changes in patients with rickets may be accompanied by impaired gastrointestinal motility and a tendency to constipation. The anterior wall of the abdomen can lead to changes in the nature of the "frog" abdomen due to muscle hypotonia. Most patients have hypochromic anemia. Individual deformities in the bones and changes in the internal organs are not always observed, and severe forms are detected. Differential diagnosis with tubulopathy (rickets) has been made in recent years due to its rarity. Mild rickets develops in only 60% of children under 1 year of age. During the convalescence, the general condition improves slightly, vegetative and neurological changes disappear, the amount of Sa and R in the deposit becomes irregular.
Children who have experienced rickets and have not been treated may have a negative impact on their subsequent growth and development. Individual deformity, scoliosis, flat heel are preserved in the bones. Occasionally, powdery mildew deformity (flat rickettsial powder) may also develop. Caries of deciduous teeth, and later permanent teeth, is often detected. In

severe cases, it even lags behind in growth. Inability to see a loved one at school age can lead to the development of myopia.

Symptoms of rickets in the period of convalescence.

Central and autonomic nervous system
Decreased sweating. Restoration of sleep. Recovery.
Muscular system
Muscle hypotension is reduced. The movement of the joints increases.
Changes in bone.
The skull is square in shape. "Olympic forehead", "saddle nose". Disorders of tooth decay. Dental caries, enamel defect. Deformation of the thorax. Change of lower aperture ("garrison owner"). Deformity of the spine (kyphosis, lordosis, scoliosis). Wrinkling of long tubular bones. "Ricky flat pelvis", "rosary" on the ribs. "Bracelet" in the wrist area. Gray fingers are pearly branches between the joints.
Laboratory indicators
Conda Sa levels are slightly reduced or normal. The amount of R in the deposit is normal or increased. At alkaline phosphatase levels. Alkalosis.
Radiological changes.
Uneven density of growth zones in bones.
Rickets level 1 is characterized by changes in the MNS, musculoskeletal system during weight gain.
Rickets is characterized by individual changes in the musculoskeletal system at grade 2 severity.
When rickets of the 3rd degree of severity is observed softening of the base of the skull, exophthalmos, nasal congestion, deformation of the Olympic forehead, thorax and spine, thinning of the epiphyseal part of the leg bones. In children with rickets, the timing and eruption sequence of teeth is disrupted. Muscle hypotension is pronounced, the size of the abdomen increases, as well as the development of static, motor functions (sitting, walking, standing) lags behind. Dysfunction of internal organs and systems is observed.
In many cases, hypochromic anemia develops, which is not the result of Fe deficiency, but is associated with structural and functional changes of erythrocyte membranes.

The process of rickets.

1. Acute - most often observed in the first half of childhood. Characteristic in this case:
- Rapid increase in symptoms
-predominance of osteomalacia process over osteoid hyperplasia.
2. Acute subacute is more common in children with intrauterine or postnatal malnutrition, in premature infants, in early infancy, in those who did not

receive adequate doses of vitamin D in the first half of life. It is characterized by:
-slow development of the disease;
-predominance of symptoms of osteoid hyperplasia over osteomalacia.
When rickets is acute, it can progress to acute exacerbation in children with acute respiratory disease.
3. In the course of recurrence - the clinical manifestations of the disease are accompanied by periods of improvement and deterioration.
4. Rickets can have a recurrent course if left untreated or not completely cured.
Clinical manifestations of rickets:
-craniotabes
-delay of healing of iliac crests and skull sutures.
protrusion of the forehead (dungpeshona)
-defect of tooth enamel
-Rach bracelets on the wrists when palpated
Appearance of -O and X-shaped legs
- "chicken" breast, "etikduz" breast.
- "garrison" owner
- "rickets" rosaries on the ribs

Treatment of rickets

Improving Sa-R metabolism for the treatment of rickets, eliminating acidosis. It aims to eliminate vitamin D deficiency. Treatment of rickets should be on a complex basis. It is necessary to organize an agenda for the child, which should be appropriate for the age of the child, and to exclude from the lights, loud noises that affect the child. It is necessary to have enough travel in the open air, regularly ventilate the rooms of the house. LFK, massage, baths and rubs play an important role. If the baby is breastfed, it is important to pay attention to the mother's feeding. If the baby is on artificial feeding, it is important to choose a formula that is close to breast milk and contains vitamin D3. In addition, vitamin D3 should be used in the treatment of rickets. In humans, the therapeutic dose of vitamin D3 is 2000-5000 XB per day for 35-40 days, then the prophylactic dose is 500x5 per day, which should be taken every day for 2 years and in the winter of the 3rd year. A new, water-soluble vitamin D3 (TERPOL, POLAND) has now been released. It has several advantages over the oil solution:
-Fast in OIT (5 times faster than usual)
-effect lasts (water-soluble vitamin D3 lasts up to 3 months after administration). Water-soluble vitamin D3 is released in drops (1 drop equals 500 XB). According to the results of the study, the effectiveness of vitamin D3 in the treatment of rickets in children is high.
The side effect of this drug is load.

In addition, in the treatment of rickets, calcium supplements are prescribed to children on natural diets and premature babies. Calcium supplements are prescribed to children in the 1st and 2nd half of life for 3 weeks. The dose of Sa glycerophosphate is 0.05 g 2-3 times a day, calcium gluconate 0.15-0.25 g 2-3 times a day. In the 2nd year of life, it is fed with calcium-rich foods. The source of Sa salts is egg yolk.

3G`1-4G`1 is mixed with a teaspoon of lemon juice and ordered in combination with 1G`4 (in water) or citrate mixture. This improves the uptake of Sa and R in the intestines and increases the reabsorption of R in the kidneys (3 times a teaspoon for 10-12 days).

In order to normalize the function of the thyroid gland and reduce autonomic disorders, potassium supplements (panangin, asparkam) are added at a rate of 10 mgGkg per day for 3-4 weeks.

In order to accelerate metabolic processes potassium-orotate 10-12mgG`kg. Carnitine-hydrochloride per day (20% aqueous solution of 4012 trigidone 3 maxal is given for 1-3 months), buy, improves weight, improves muscle hypotension, metabolic processes.

In children with severe rickets, 0.5 mg of ATF is given intramuscularly for 15-30 days. After a comprehensive drug therapy, sung LFK and massage are prescribed. Sung baths are taken 1 month after the start of therapy. Mild baths are recommended for children (1 liter of extract 360S DA per 10 liters of water, the first bath is 5 minutes, the next 8-10 minutes, 13-15 treatments). Salt baths are mainly for weak, sedentary children (2 tablespoons of sea or table salt are added to 10 liters of water, the first bath 3 minutes, the next 5 minutes 8-10 treatments). Hot baths (sachratki, air root, chamomile, etc.) are ordered. Treatment with baths is carried out 2–3 times a year. In severe cases under the supervision of the dispensary they stay for 3 years. They should be on dry land every quarter. Specific prophylaxis is carried out for 2 years in autumn and winter, and 3 years only in winter. Rickets cannot be a prophylactic contraindication. Vitamin D3 is vaccinated after 2-3 weeks.

Prevention of rickets

1.Antenatal (during the fetal period) it can be specific and nonspecific. Nonspecific prophylaxis is performed long before birth. A pregnant woman should follow the agenda. Day and night sleep should be adequate. It is important to walk in the fresh air for 2-4 hours a day, to eat rationally: 180-200 g of meat, 100 g of fish, 150 g of cottage cheese, 30 g of cheese, 500 ml of milk or cottage cheese, as well as foods rich in micronutrients and macronutrients. should be fed with.

Pregnant women at risk (nephropathy, diabetes, hypertension, rheumatism) are prescribed additional vitamin D 1000-1500 XB per day for 28 weeks at 28-32 weeks of pregnancy, regardless of the season. In winter and spring, and in the northern regions, 1-2 courses of UFO instead of vitamin D

throughout the year (starting from 1.4 biodoses, gradually increasing to 2.5-3 doses). UFO promotes endogenous synthesis of cholecalciferol.

2. One of the main measures for postpartum childbirth is the organization of proper nutrition. Babies under 2 months of age are best fed with this breast milk. A breastfeeding woman should follow a routine and eat properly. Multivitamin preparations, dairy products should be consumed regularly. In cases not fed with breast milk, adapted breast milk substitutes, ie vitamin D3-containing mixtures, are prescribed.

   Great attention should be paid to the physical development and hardening of the child: it is a walk in the fresh air, exercise therapy, massage, hydrotherapy, carried out systematically: regularly, for a long time, gradually.

   Specific prophylaxis is carried out with vitamin D, prophylactic clean 400-500 XB per day for a healthy newborn, it is given from 3-4 weeks, in the autumn-winter-spring months, and the child's living conditions and risk group are assigned to sick children. Specific prophylaxis is prescribed to infants in the autumn-winter-spring months in the 1st and 2nd year. Children at risk of rickets are prescribed daily vitamin D 1000XB in autumn-winter-spring for 2 years.

   Specific prophylactic doses of rickets are prescribed to first-born premature babies from 10-14 days of life 400-500XB per day for 2 years, except summer. Premature infants of grade 2-3 are prescribed 1000-2000XB per day for the first year of life, and 500-1000XB per day for 2 years outside the summer months. Specific prophylaxis of rickets in an aqueous solution of vitamin D3 is preferable, especially for premature infants, as they have a lack of enzymatic activity in the intestines. Contraindications to the prevention of vitamin D3 may be:

   -Idiopathic calcification.

   -Hypophosphatism.

   -Organic lesions of the CNS, with symptoms of microcephaly and craniostenosis.

   Children with small amounts of Likildok have contraindications to vitamin D prescribing. They are given specific prevention of rickets from 3-4 months of life

Criteria for assessing the knowledge and skills of groups.

86 – 100 score	The student has a thorough knowledge of inherited phosphate diabetes, tonidebrifanconi, Ellers-Danlo and diseases associated with connective tissue metabolism disorders and knows their etiology, clinic, diagnosis and principles of treatment. Can differentiate them from each other.
71 – 85 score	The student knows the etiology, clinic, diagnosis and treatment of diseases associated with inherited

	phosphate diabetes, tonidebrifanconi, Ellers-Danlo and connective tissue metabolism disorders. Can differentiate them from each other
55 – 70 score	Responds moderately to questions on the subject, does not fully understand the etiology, clinic, diagnosis and principles of treatment of diseases associated with inherited phosphate diabetes, tonidebrifanconi, Ellers-Danlo and connective tissue metabolism disorders. Cannot differentiate them from each other
0 – 55 score	Hereditary phosphate has a shallow knowledge of diabetes, tonidebrifanconi, Ellers-Danlo disease. Cannot differentiate them from each other.

PRACTICAL LESSON 5

Multifactorial diseases

Class time - 2 hours	Number of students: 18
Class type	Practical training
Plan	The role of genetic and environmental factors in the origin of multifactorial diseases, diagnosis, treatment, prophylaxis, birth defects, ischemic heart disease, peptic ulcer disease, rheumatoid arthritis, diabetes, schizophrenia, cholecystitis, urinary tract stones, oncological, allergic diseases.
The purpose of the topic: - Know how to develop risk factors, - timely diagnosis, - Methods of testing and identification of risk factors in young families - To teach to use easy and convenient methods of detection of infertility in couples.	
Task of the topic - - Formation of interest in the profession, humanity, a sense of responsibility; -Education of interest in expanding their knowledge, development of students' thinking skills, logical hinking; - Guide students to a creative approach to the study of the topic.	
Teaching methods	Demonstration, lecture, conversation

Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

1.2.Practical Lesson Technology Map.

Stages and timing of work	Educator	Learners
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment 5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They are divided into small groups They watch They participate They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Questions on the topic:

1. Describe the clinic, pathogenesis and treatment of multifactorial disease etiology
2. Describe the clinic, pathogenesis and treatment of Down syndrome etiology
3. Describe the etiology, pathogenesis and treatment measures in Patau syndrome
4. Shershevsky - to describe the clinic of etiology, pathogenesis and treatment of Turner syndrome
5. Describe the clinic, pathogenesis and treatment of the etiology of Klinefelter's syndrome
6. Describe the clinic, pathogenesis and treatment of the etiology of cat scream syndrome

Interactive method “Wheel”.

Technology of interactive methods used in training:

Description of technology. this interactive method teaches students to think logically about the practical skills they have learned as a result of the topics covered, to answer the questions independently and correctly, and to self-evaluate, and in a short time by the teacher. aimed at assessing the knowledge acquired by all students.

The purpose of technology is to teach students to think logically in the classroom, to express themselves freely, to evaluate themselves, to work individually and in groups, to respect the opinions of others, to choose from many ideas.
Tools used in the lesson:
handouts (Situational and problem tables)
colored pencils (or felt-tip pens).
Schedule:
grouping students (depending on the circumstances);
to acquaint students with the requirements and rules of the course;
distribution of handouts to group members.
the members of the group independently perform the tasks in the handouts;
Each group member writes the group number in the right corner of the handout they have worked on, and draws any of his or her own symbols in the left corner;
handouts are exchanged for other groups in the direction of "wheel rotation";
materials submitted by new team members will be reviewed and amended;
The game continues until the answers are under the control of all groups
Each group will then receive a piece of paper with their mark on it and the ideas expressed will be discussed.

Topic content

Gastric or peptic ulcer: The symptoms of peptic ulcer disease are different. Classical symptoms may not always be observed: severe pain in the epigastric area observed 1-3 hours after a meal; nocturnal pain in the epigastric area, causing patients to wake up early in the morning in duodenal ulcers. Transmission of pain to the back of the body may be a sign of posterior wall penetration of duodenal wounds. H. pylori gastritis is detected in 80-90% of cases of 12-finger ulcers and 60-70% of cases of peptic ulcers. The high risk of developing peptic ulcer is inextricably linked to certain medications (especially aspirin and other NYaQV corticosteroids), the presence of ulcer disease in family members, smoking, alcohol consumption, and stressful situations. 12-finger ulcer disease is 4 times more common than peptic ulcer disease. Laboratory analyzes. A complete blood count, a latent blood stool test (Gregersen reaction) is performed at a single diagnosis or in cases of dangerous exacerbation of the disease. Bleeding from the upper parts of the gastrointestinal tract is detected in 25% of cases. It is observed as an initial sign in 10% of patients. Perforation develops in approximately 5% of cases, especially in patients receiving NYaQV. The diagnosis of ulcer disease is made on the basis of the results of endoscopic or radiological (with barium) gastroduodenal examination. Treatment without drugs Feed in small portions 4-5 times a day. Exclude spicy, salty, smoked products from the diet, as these products can cause symptoms. Stop consuming alcohol and smoking. Discontinue taking the following medications: aspirin, NYaQV, corticosteroids, reserpine. Drug treatment Treatment of ulcer disease explained

by H. pylori is performed using a three- or four-component treatment regimen. Three-component treatment scheme. Omeprazole 20 mg orally, every 12 hours, for 10-14 days. Amoxicillin 500 mg orally, 4 times a day or 1000 mg 2 times a day for 5 days. It is recommended to take Metronidazole 500 mg 2 times a day for 7 days after meals. The four-component treatment regimen includes three-component treatment drugs and bismuth colloidal subcitrate (De-Nol) 120 mg 3 times a day 30 minutes before meals and a fourth before bedtime for 10-14 days. Antacids. Treatment is continued for 4-8 weeks: it is recommended to take ranitidine 300 mg or famotidine 40 mg at 8 pm or omeprazole 20 mg every 12 hours. Treatment of wound disease associated with the reception of NYAQVs. Stop taking NYAQVs. Prescribing antacids: ranitidine 300 mg or famotidine 40 mg at 8 hours or omeprazole 20 mg every 12 hours, recommended for 4-8 weeks. If it is not possible to stop taking NYAQVs, omeprazole or another proton pump inhibitor is prescribed. Criteria for referral Patients should be referred to the hospital immediately in the following cases: if there are signs of bleeding, penetration or perforation; acute pain or frequent recurrences; presence of wounds of one or more large sizes (3 cm and larger); if symptoms persist for 7 days despite adequate treatment; severe manifestations of diseases of the gastrointestinal tract (chronic hepatitis, liver cirrhosis, chronic recurrent pancreatitis). Complications of Wound Disease Complications of Wound Disease (bleeding, penetration, perforation, stenosis, malignancy) may be an indication for surgical intervention. Bleeding is a common complication.

The word psoriasis is Greek and means itching. Among the common people, the disease is also known as scabies. It is a severe and persistent pain that belongs to the group of skin diseases that occur in the form of balls or rashes.

In place of information…

Psoriasis is not only a skin disease but also a general disease of the body and affects the internal organs. The disease affects the nervous system, endocrine system and liver.

By the way…

The disease also depends on the weather. During the summer months, some patients develop psoriasis completely. In some cases, exposure to sunlight can cause the disease. The third group of patients develops the disease in spring and autumn.

Factors contributing to the disease:

• Infectious diseases (influenza, acute respiratory viral diseases, chickenpox, mumps, hepatitis);
• Pneumonia, pyelonephritis, etc .;
• purulent otitis, chronic tonsillitis, sinusitis, frontitis, dental caries, etc .;
• strong fear and excitement;
• stresses;
• depression;
• some medicines;
• dietary disorders and b.

Symptoms of the disease

There are pink, inflamed spots on the skin. Rash can also be observed on the hairy part of the head. The skin around the spots is a little red. Then the stain gradually gets bigger. The spots tend to stick together. The spots are usually symmetrical and occur on the outer surface of the limbs. Periodically forms folds. These spots are most often found on the most friction areas of the skin. For example: facial skin, genital area, around natural wrinkles, buttocks.

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Factors contributing to this disease:

Fried, fatty foods, kazi, kebabs, eggs, milk, confectionery, store sweets, coffee, cocoa;

Even when these products are consumed in moderation, their energy is wasted or consumed in very small quantities. Over the years, the body's energy levels increase. This condition gradually warms the blood and causes disease.

• hereditary predisposition;
• diseases of internal organs;
• climatic conditions;
• metabolic disorders, etc.

Types of diseases

1. A type of rash that spreads all over the human body.
2. A bulge in some parts of the body.

Scabies can be hereditary. But malnutrition is often the cause of illness. The disease is often chronic and recurrent. In children, the disease often recurs during the winter months.

By the way…

Scabies can affect people of all ages. But it usually starts after the age of 50.

- Proper nutrition is an important factor in the treatment of kidney stones. If the kidney has urate salt stones, the patient should eat foods consisting of dairy and plant products. When phosphate salts form, it is better to eat meat. In this case, dairy and vegetable products should be avoided. It is also strictly

forbidden for people with kidney stones to eat beans, peas, moss, peppers, fried and smoked foods.
- In the treatment of pyelonephritis it is necessary to eat only as prescribed by a doctor, and most importantly to consume less salt.
- Patients with nephritis should beware of heavy physical labor, chills and nervousness. In addition, such patients should be under medical supervision at the dispensary, to avoid infectious diseases.
- People with kidney disease, such as urinary stones, nephritis, pyelonephritis, should exercise and be very careful about infectious diseases such as tonsillitis, pharyngitis, angina, which are caused by streptococci.

QUACK DOCTORS' TREATMENT

- Cooked garlic, eaten with radish seeds, helps to reduce kidney stones.
- If you crush a pea and drink a thick infusion in a teapot, the stones that appear in the kidneys and bladder will fall off.
- One hundred grams of parsley and one large flood of finely chopped dill root, add to it one hundred grams of honey and a liter of water, then stir until boiling over low heat. It is allowed to rest for three days. Then add another liter of water and bring to a boil. Strain through a sieve without cooling. If you drink three tablespoons before a meal for ten days, the sand in the kidneys will be washed away.
- Patients with inflammation of the kidneys benefit from drinking pumpkin seeds. Also, squeeze the flesh of the pumpkin raw and drink half a glass a day. This treatment is useful when there is sand, stones in the bladder and kidneys, corrects inflammation there.
- Vitamin K deficiency can also lead to kidney disease. Therefore, it is necessary to consume products containing vitamin K (phylloquinone) (spinach, cabbage, green part of nettle).

Kidney diseases

The main symptoms of these diseases are swelling of the body, high blood pressure (hypertension), changes in the composition of urine, the formation of protein, erythrocytes, leukocytes. Acute inflammatory diseases of the renal parenchyma - diseases that affect the ball and tubular apparatus of the nephron, causing tumors in the body.

Tumors appear quickly, often starting on the face and spreading to the limbs. Occasionally there is a build-up of water in the cavities. The skin at the site of the tumor is pale. In terms of the development of tumors associated with the kidneys, it is associated with impaired capillary permeability and a decrease in the amount of protein in the blood plasma. Elevated blood pressure in kidney disease is associated with impaired renal hemodynamics and ischemia of renal tissue.

Hypertension continues with the onset of excruciating headaches, dramatically increasing the load on the left ventricle of the heart and can lead to cardiac asthma. In kidney disease, it is important to check the urine, which can be used to determine the amount of urine per day, the reaction of the relative density of

urine, the presence or absence of proteins, the composition of urine sediment. Most kidney diseases are accompanied by proteinuria (albuminuria) in the urine and the appearance of cylinders as a result. In acute inflammation of the renal parenchyma, tumors of the kidneys and urinary tract, in various diseases of the mucous membranes of the bladder, urinary tract, the appearance of blood clots in the urine, hematuria , the blood sometimes enters the urine in the form of a stream of flesh (macrohematuria) or the urine sediment becomes visible when examined under a microscope (microhematuria).

Inflammatory diseases of the urinary tract, kidney vessels, bladder continue with the appearance of leukocytes in the urine, pus (pyuria). Changes in the rhythm of bladder emptying, an increase in the amount of urine excreted at night (nocturia), as well as a decrease in the relative density of each batch of urine detected by the Zemnitsky test (q) indicate impaired renal concentration function will give. In the later stages of kidney disease, the kidney's ability to excrete nitrogen is usually impaired, and this can be confirmed by examining the patient's venous blood for residual nitrogen. Normally, the amount of residual nitrogen in the blood is 14.3 - 28.6 mmol / l. Increased residual nitrogen in the blood due to impaired renal nitrogen excretion (azotemia) leads to a gradual intoxication of the body with the products of protein metabolism and the appearance of a clinical picture of uremia. Nitrogenous wastes are excreted by the gastrointestinal tract to compensate for the lack of renal function, but nitrogenous wastes cause the mucous membranes of the stomach and intestines to be affected, causing the patient to vomit and have diarrhea. will be. Acute diseases of the renal parenchyma can sometimes lead to circulatory disorders in the brain, nausea, as well as headaches and high blood pressure.

A patient with acute or severe chronic kidney disease should be hospitalized. He must lie down without getting up. A person's warm horizontal position allows the renal vessels to dilate and improve intrarenal hemodynamics. In addition, lying down reduces the body's energy expenditure and the load on the damaged kidney tissue. The nurse is very attentive to the patient lying down, explaining to the patient that violation of this rule, excessive physical activity can lead to an acute form of the disease, which can lead to an incurable chronic form.

To improve renal hemodynamics, dry heat in the form of warm-ups is applied to the lumbar region. When there is a sharp decrease in urine output (oliguria), deatermia is prescribed to the lumbar region or paranephral blockade with novocaine by the Vishnyovsky method. Both of these treatments improve blood circulation in the kidneys and increase diuresis. During a paranephral blockade, the nurse looks at the doctor, prepares instruments and medications, and looks at the patient's condition after the blockade.

Adherence to a diet is one of the main methods of treating kidney disease. The nurse monitors the patient's adherence to the prescribed eating and drinking regimen. In acute renal disease, which is accompanied by severe swelling of the skin and hypertension, it is recommended not to eat or drink for 1 to 3

days, or to limit fluids and give glucose with wet fruits. In the first case, the patient does not eat or drink anything, and glucose is sent intravenously along with vitamins. Such a strict diet usually leads to improved mood, the disappearance of tumors in the body, a decrease in blood pressure, after which the diet is gradually expanded, strictly limiting the amount of salt to 3-4 g per day. Any smoked, salted or seasoned ingredients are prohibited. Dietary №7 and №7b meet such a requirement. Fresh fruits and vegetables should be given plenty of vitamin C. To enhance diuresis, a diet that facilitates - sugar, fruit watermelon days are prescribed. The nurse explains the procedure to the patient. Potassium salts help to reduce swelling in the body and the forced excretion of fluid from the body, which is given in the form of drugs and potassium-rich foods (rice, leaves, potatoes).

After the suppression of acute events and in chronic kidney disease, when the nitrogen-fixing function of the kidneys is preserved, the content of dietary proteins, carbohydrates, fats and vitamins should be stable, but the salt content should be up to 5 g per day. should be limited; salty foods, alcohol are strictly prohibited. In cases of impaired renal nitrogen excretion, the daily protein intake is sharply limited, and in some cases, a completely protein-free diet. It is advisable to give patients plenty of fluids and parenteral fluids. All this reduces the toxicity of the organism with nitrogenous wastes, products of protein metabolism. In renal disease, which is accompanied by severe albuminuria and, consequently, a decrease in the amount of protein in the blood plasma (onset of hypoproteinemia), it is recommended to give a large amount of complete protein with food, while the patient is given 2 g of protein per 1 kg of body weight. . Patients are additionally prescribed boiled meat, fish and cottage cheese. Drinking fluids and salt are limited. Such patients are sometimes given blood plasma.

Patients with acute kidney disease or chronic disease should be closely monitored and cared for. seemingly insignificant changes in the patient’s mood and condition can be signs of horrible complications and should therefore not be overlooked by the nurse. Excessive dryness and itching of the skin, loss of appetite, nausea, vomiting, and increased thirst are signs of worsening kidney failure and the onset of uremia. It is also important to monitor the psyche of patients, as azotemic uremia often begins when patients are relaxed, drowsy during the day and sleep deprived at night, and have blurred vision. If you have any of these symptoms, tell your doctor right away.

Most of the acute and chronic kidney diseases are accompanied by heart failure, in which case the nurse must monitor the patient's breathing, pulse rate, blood pressure. The nurse will report the results to the attending physician. In acute cases of kidney disease, most emergency procedures include blood transfusions, intravenous infusions, gastric lavage, and topical cleansing enemas. In order to provide timely care to the patient, the nurse must carefully study these procedures.

Kidney stone disease

Kidney stones due to impaired mineral and protein metabolism. Urinary tract infections cause stones to form, which prevent the flow of urine, elongation of the renal vessels, and then compression of the renal parenchyma. Urinary incontinence, in turn, can lead to an increase in urinary tract infections. Stones can damage the mucous membranes of the urinary tract with kidney stones and cause hematuria. The disease is manifested by a sudden seizure of the kidney, which is prevented by the stone being pushed along the urinary tract. The time elapsed between one fork and a second fork can vary widely. When kidney stones are not a complication, the patient usually feels completely satisfied when the kidney stones do not go away. Renal colic is characterized by the sudden onset of severe pain in the lumbar region, which is on the patient's side. Intermediate, genitals, legs. As long as it stays poisonous, it goes away. Vomiting is often accompanied by reflective vomiting.

The pain is unbearable in nature, the patient finds no place for himself and is thrown everywhere, lying on the bed from side to side. Micro and macrohematuria usually occur during a kidney attack. If renal colic cannot be treated at the patient's bedside, the patient should be admitted to the urology department of the hospital.

In addition to antispasmodics and narcotic analgesics, it is beneficial to place hot tubs in the lumbar region and place the patient in a hot bath, which eliminates urinary tract spasms and thus helps the stone to pass. The nurse obeys all the doctor's orders and does not leave the patient until the pain is relieved. Urine should be checked after a fork. Treatment for colic should be aimed at restoring metabolism and preventing the formation of new stones. For this purpose, an appropriate diet is prescribed and plenty of fluids are drunk; treatment of urinary tract infections is prescribed.

The nature of the diet depends on the chemical composition of the stones. In the presence of urate salts, it is necessary to limit the consumption of meat and extractives (coffee, cocoa, broth and mushroom decoctions). Calcium-rich foods (dairy products, eggs, pastries) are not recommended in the presence of phosphate stones. Meat, fish and pastries can be eaten instead. Drink at least 2 liters of fluid every day.

Pyelitis is an inflammation of the kidneys. It is caused by the passage of microorganisms into the kidneys. The disease is most often caused by Escherichia coli. Due to the presence of a mechanical barrier (stone, prostate adenoma, enlarged uterus during pregnancy), stagnation of urine in the renal pelvis leads to the appearance of honey. The disease is characterized by a sudden onset of fever with a rise in temperature to 30 - 40 C, with the appearance of throbbing pain in the lumbar region. Most people are frequently poisoned and irritated when urinating. The urine is cloudy.

Examination of the sediment under a microscope reveals a large number of leukocytes. Patients are often successfully treated at home. When the temperature rises, a person should not lie down. It is recommended to drink plenty of fluids, the amount of salt should be limited. In acute cases, the use of

antimicrobials (sulfonamides, broad-spectrum antibiotics, furadonin) is beneficial.
Chronic nephritis is often the result of a previous acute nephritis. The acute phase of the disease sometimes goes unnoticed by the patient and others, and it is only after the onset of the clinical picture of true chronic nephritis that the acute nephritis becomes known. Chronic nephritis exacerbates from time to time, exacerbation of the disease leads to an increase in the number of balls and the onset of renal failure and azotemic uremia (death of patients with chronic nephritis is usually caused by azotemic uremia) .
The Zimnidsky test showed that the relative density of urine was low, varied slightly in different portions, the number of nocturnal urinary portions was increasing, the renal parenchyma was more damaged, many balls were damaged, and connective tissue was replaced. indicates that it is appearing. Outbreaks appear to be exacerbated by streptococcal infections (angina, erythema multiforme), viral infections of the upper respiratory tract, and chills. therefore, a patient with chronic nephritis should beware of colds, cold sores, and wear warm clothing and shoes during the cold seasons of the year. Work should not be associated with physical exertion or exposure to cold.

In acute nephritis - urinary syndrome - characterized by proteinuria, cylindrical, macro and microhematuria. Not only finely dispersed albumins but also globulins and fibrinogen are released through the capillary walls of the injured balls. The amount of protein in the urine ranges from 1 to 10%, in some cases up to 20%. But protein in the urine only increases during the first 7-10 days. Therefore, in late tests, the level of albumin in the urine is not very high (less than 1%). Minor proteinuria sometimes occurs at the onset of the disease, and in other cases, may not occur at all. Low levels of protein in the urine can persist for a long time in acute nephritis. This condition usually disappears in 3-4-6, sometimes 9-12 months. Hematuria is a persistent and inevitable symptom of acute glo-merulonephritis. In 13-15% of cases, macrohematuria is detected, in other cases, microhematuria is detected, in which the number of erythrocytes in the urine sediment does not exceed 10-15 in the field of view.
In acute glomerulonephritis, cylindrical may not be observed. Only 75% of patients have hyaline, granular, sometimes epithelial cylinders. White blood cells are a rare symptom of glomerulonephritis, in which the number of leukocytes is lower than in erythrocytes. Deficiency of leukocytes from erythrocytes is determined by examination of urine sediment by Kakovsky-Addis, Nechiporenko methods.
Decreased kidney function can sometimes lead to low levels of nitrogen in the blood and an increase in urea. Azotemia does not last long.
In acute nephritis, there is a decrease in hemoglobin and erythrocytes in the blood.
Anemia is caused by hydremia (a blood clot) and an infection that causes nephritis (such as septic endocarditis).

The rate of erythrocyte sedimentation rate increases sharply, with swelling, shortness of breath, headache, back pain, and decreased urine output.
fat, protein and blood in the urine, and high blood pressure. The tumor is observed for 2-3 weeks, then the amount of urine increases (polyuria), blood pressure drops, the specific gravity of urine decreases, but proteinuria, microhematuria may last longer.
The second type of acute nephritis, latent type, often progresses to chronic nephritis. In this type, the disease begins slowly, with a slight shortness of breath and swelling in the legs. Consecutive tests show changes in the composition of the urine, which can last for 2-3 months.
Acute nephritis can lead to renal eclampsia. Eclampsia develops when there is an increase in blood pressure and excessive swelling. In renal eclampsia, the patient may have weak eyes, frequent muscle spasms, narrowed arteries, fainting, bite of the tongue, and involuntary stool and urinary incontinence. This condition resolves after medication.
Another serious complication of acute glomerulonephritis is nephrotic syndrome, which is characterized by excessive protein loss, extremely severe tumors, decreased protein in the blood, dysproteinemia, and increased cholesterol (hypercholesterolemia). ¬nadi. The pathogenesis of nephrotic syndrome is based on a widely accepted immunological concept. Nephrotic syndrome is called by sending a nephrotoxin serum to form in the experiment. Disruption of the renal basal membrane plays an important role in the development of nephrotic syndrome. Antibodies to kidney proteins accumulate in the basal membrane and cause a constant complementary reaction. When nephrotic syndrome progresses, the complement in the blood decreases. Antibodies to the kidneys increase. Immunosuppressive drugs The positive effect of steroid hormone on nephritis confirms that the disease has immunoallergic properties.
The second hypothesis explains the development of nephrotic syndrome
is the concept of metabolic disorders. According to this concept, the combination of protein with urine is due to a violation of the filtering properties of the renal glomeruli. Modern studies have shown that in nephrotic syndrome there is an increase in the activity of soluble proteins in the kidneys and urine, an increase in histamine in the blood, high concentrations of lysosomal enzymes in the kidneys, which cause their inflammatory response. Disruption of protein balance in serum leads to the release of their finely dispersed protein fractions and immunological shifts in the body.
Other data suggest that impaired protein synthesis in nephrotic syndrome is caused by changes in the function of the reticuloendothelial system. In nephrotic syndrome, the increase in fat in the blood is compensatory, which is inversely proportional to the decrease in protein in the blood. The pathogenesis of tumors should not be associated with a decrease in blood protein and a decrease in colloid-osmotic pressure in plasma. In nephrotic syndrome, sodium retention, increased reabsorption, and secondary hyperaldosteronism are associated with increased aldosterone production,

which in turn leads to circulatory disorders. Not only increased permeability of the renal capillaries, but also immunological damage to the capillaries in other tissues is a factor in nephrotic syndrome.

From a clinical point of view, the most important pathogenesis is protein excretion in the urine, which is observed in nephrotic syndrome up to 3 g / day or more. The formation of protein in the urine is mainly due to damage to the basal membrane of the glomeruli and increased permeability, as well as the entry of large protein molecules through the glomerular filter. It should also be noted that the normal function of phagocytes prevents the entry of protein molecules in the basement membranes. Impaired function of podocytes

As a result, proteins cannot be retained and they enter through the basement membrane. Any acute nephritis that goes unnoticed throughout the year can lead to chronic nephritis. It should also be noted that acute-onset diffuse glomeralonephritis sometimes results in moderately acute extracapillary nephritis, and its poorly progressive course can lead to chronic renal failure in the coming months.

Diagnosis. Acute diffuse glomeralonephritis is not difficult to diagnose because the disease occurs in young people and begins with obvious clinical signs. A comparative diagnosis between acute nephritis and recurrence of chronic nephritis is difficult. It takes 1-2 weeks from the onset of the disease, from infection (tonsillitis, angina, rhinitis, etc.) to acute nephritis. Chronic glomeralonephritis is characterized by high blood pressure, left ventricular hypertrophy, and marked damage to the fundus.

Changes in urine may be different, but chronic nephritis is characterized by a decrease in urine density and filtration function (hypo-, isostenuria), acute nephritis is characterized by heart failure (shortness of breath, edema, asthma, bradycardia), disease o Acute origin (macro- and microhematuria). The comparative diagnosis of latent acute nephritis and pyelonephritis is difficult. This is due to the predominance of erythrocytes in the urine sediment over leukocytes, the absence of proteins, the absence of anamnestic indications, X-ray, ultrasound, scan, radioisotope, biopsy. A comparative diagnosis should be made between acute nephritis and renal tuberculosis and other kidney diseases.

Treatment. The treatment of this disease requires careful care of the patient, adherence to the treatment regimen and diet. The role of the nurse in this process is great. Great care should be taken with skin care (i.e., swelling, bed sores), bowel function, and heart rate should be monitored regularly. Diuresis from urine relative to drinking fluid

It is necessary to calculate not only the amount of urine, but also its color and clarity in relation to the fluid that is excreted from the body (sweat, vomiting, diarrhea). It is necessary to lie still, diet, take medication, avoid complications and infectious diseases until the swelling disappears and blood pressure returns to normal (3-4 weeks). The patient was given a low-salt diet (Table 7), antibiotics (penicillin, ampicillin) for 10-14 days; from sulfa-nilamides: nitroxoline, polyne gramurine and others are prescribed. Antiallergic, desensitizing drugs (diphenhydramine, calcium chloride) are useful, high

blood pressure, hypotensive drugs dibazol, papaverine, adelfan, clofelin, raunatin, rauvazan, apressin are recommended. Diuretics such as lasix, furosemide, hypothiazide, verospirone, anticoagulants, and antiplatelet agents are also prescribed.

In nephrotic syndrome of acute glomerulonephritis and its prolonged cases, corticosteroid hormone pred-nizolon 60-100 mg per day for 4-8 weeks, then gradually reduced, diathermy from physiotherapy treatments lowers blood pressure and relieves back pain, reduces swelling. Discharged patients remain in outpatient care for up to 4 months until complete recovery (even when acute glomerulonephritis is well).

Consequences. Acute nephritis is completely cured. Acute nephritis kills. It occurs only in cerebral hemorrhage, heart failure, pneumonia. Acute nephritis is chronic in 1/3 of cases. The outcome of acute nephritis depends to some extent on the early detection and proper treatment of the disease. Currently, the use of corticosteroids has significantly reduced the incidence of the disease. In the acute phase, patients lose their ability to work and need to be hospitalized. It usually heals in 2-3 months. Patients can return to work. Patients with acute nephritis should be monitored at the dispensary, even if there is urinary syndrome and residual albumin. This is especially important for patients with urinary syndrome. Irreversibility of the disease

to prevent infectious lesions in the body. The patient should avoid year-round exposure to cold, especially wet, cold.

Prophylaxis. Patients with acute glomeralonephritis are registered and monitored by a nurse every 10 days after discharge, then once a month, and then once every 2-3 months. Patients with this disease should not work in cold and humid rooms, and should not engage in strenuous work. It is not possible for a woman with this disease to become pregnant for up to 3 years. The patient is sent to spas with dry and hot climates. Infectious diseases and foci of infection should be treated, the oral cavity should be rehabilitated, and cooling should be avoided.

5.1.1. Chronic gross glomeralonephritis

Chronic glomeralonephritis is a long-lasting immunoallergic inflammation of the glomeruli of the kidneys. The disease is characterized by a decrease in renal function, increased arterial blood pressure and the development of renal failure due to gradual injury of the renal glomeruli.

Reasons. Chronic nephritis develops after acute glomerulonephritis and also as primary chronic glomerulonephritis. Chronic glomeralonephritis is mostly immune in nature and develops under the influence of serums, vaccines, antigens, toxins, some medications, and colds. Some drugs can cause not only "acute" toxic kidney disease, but also the development of generalized glomerulonephritis. Chronic glomeralonephritis also develops in tuberculosis, ulcers, bacterial endocarditis, hemorrhagic vasculitis, nodular periarteritis and other diseases.

Pathogenesis. The chronic appearance of nephritis depends on the degree of immunological, especially autoimmune, changes in the macroorganism.

Pathological anatomy. Clinical manifestations of chronic glomeralonephritis and anatomical changes in the kidneys are interrelated

If the syndrome is predominant, the kidney is large, the epidermis is easily detached, the surface is smooth, light gray (large white kidney). The cortex is an enlarged, light gray, reddish-brown part of the brain. The microscope shows enlarged iliac tubes of the kidney, edema in the epithelial tissue, wrinkles, granular or vacuolar dystrophy.

Examination of the kidneys by puncture biopsy reveals the following histological types of chronic glomerulonephritis: 1) membranous glomerulonephritis - characterized by fusion and thickening of tissues in the basal membrane of the renal glomeruli; 2) in mesangial glomerulonephritis with increased tissue density, there are changes in the tissue elements of the glomeruli; 3) connective tissue develops in the balls (fibroplastic glomeruloenephritis).

In the final stages of glomerulonephritis, the kidneys do not shrink, they shrink, the brain is covered with granules, and the cortex becomes thinner.

The vesicles loosen and are replaced by connective tissue, the kidneys do not wrinkle, and secondary kidney wrinkles appear.

Clinical landscape. The clinical picture of chronic glomerulonephritis depends on its type. The disease is characterized by three main groups of syndromes. 1. Changes in urine: proteinuria, hematuria, cylindruria. 2. Changes caused by high blood pressure. 3. Changes in the body caused by tumors. The following clinical types of chronic glomerulonephritis are distinguished.

1. Latent glomerulonephritis is the most common type, accounting for 44%. This type of glomerulonephritis is characterized by slight changes in the urine, no increase in blood pressure, and the tumor is not noticeable. The disease can reappear in 30-40 years. Patients do not lose their ability to work for many years and do not feel sick. The most common type of chronic glomerulonephritis is latent or emergency

changes in urine during a visit to another doctor or dispensary (decreased weight, more red blood cells, more urine at night), a slight increase in blood pressure, accelerated ECHT, protein in the blood decreased, is determined on the basis of increased cholesterol. Most latent suranal glomeralonephritis is diagnosed when renal function is impaired and insufficiency develops. Renal insufficiency is caused by an increase in the amount of residual nitrogen and urea in the blood.

2. Nephrotic manifestations of chronic glomerulonephritis. Through urine
large amounts of protein (more than 3.5 g per day) in the blood
protein (hypoproteinemia), especially albumin / globulin
decrease in the ratio (dysproteinemia), cholesterol mod-
increase in blood pressure (hypercholesterolemia - 600 - 800 mg%)
represented by Hypoproteinemic in the body of patients
(as a result of protein depletion) tumors appear.
Such
tumors are different from other tumors of the face and eyelids

begins and gradually spreads throughout the body. They are constant
even the internal organs, heart, lungs and
spreads to the peritoneum, cavities. Skin mucous membranes
dries, muscles shrink (atrophy), resulting in skin
does not wrinkle.
As protein is excreted in the urine, it increases the amount of triglycerides and free cholesterol in the blood.
The nephrotic form of succulent glomerulonephritis is accompanied by symptoms of nephrotic syndrome and nephritis (bleeding in the urine, decreased renal filtration).
Blood pressure may not increase in the early stages of the disease, but may increase in the later stages.
3. Hypertensive manifestations of chronic glomerulonephritis. Co-
pincha is characteristic of latent glomerulonephritis. Patients
The test usually shows an increase in blood pressure. Blood
pressure is not constant at the onset of the disease, for the most part
increases towards the evening under the influence of cold and nervous disorders. Kidney
when activity decreases, systolic and diastolic pressure increases steadily, reaching 200/120 mm Hg, gradually cardiac activity is also impaired, heart rate increases, I tone decreases in the apex area, systolic murmur occurs in the aorta Tone II increases, heart sounds are heard as fast and loud as when riding a horse, then heart failure is added, asthma attacks and even lung tumors can occur. In chronic glomerulonephritis, blood pressure gradually increases, ischemic heart disease, angina develops. There are changes in the blood vessels in the fundus of the eye, and there is a phenomenon of hemorrhage in the fundus. Only in the last stages of the disease can serious and irreversible changes in the retina occur. Blood pressure rises as the kidneys bleed and renin increases. Blood pressure is very high and stable during kidney failure and when the disease is severe. The patient may have a myocardial infarction and cerebral hemorrhage.
Mixed form of chronic glomerulonephritis. This condition is characterized by high blood pressure and nephrotic syndrome. However, the symptoms of these syndromes may not occur to the same extent. 1 or 2 syndromes occur during the peak period of most disease. For example, in the syndrome of changes in the urine (proteinuria, hematuria, cylindruria), the patient's general condition does not change for 2-5 years, is satisfactory, and then develops renal insufficiency.
The clinical course of chronic glomerulonephritis consists of 2 stages: 1. Nitrogen excretory activity of the kidneys is preserved. This stage is long, latent, and is manifested only by changes in the urine. 2. The stage of impairment of renal function. This stage can last from 1 month to 30 years after the onset of the disease. In the acute phase of the disease begins faster, in the latent phase. At this stage, kidney function decreases. Nitrogen excretion, urine thickening activity decreases. Urine changes are reduced, the specific

gravity is reduced to around 1007-1008. Protein is constantly excreted in the urine. Blood pressure rises regularly. Swelling of the skin, an increase in urea, creatinine, and protein in the blood, and a decrease in protein.

Diagnosis and comparative diagnosis. It is not difficult to diagnose if the patient has a history of acute nephritis and the clinical picture of chronic nephritis is clear. However, it is difficult to diagnose if the nephritis is latent or if the hypertensive suran nephritis is present.

If the patient does not have acute nephritis, but some protein and hematuria are found in the urine, then other kidney diseases (pyelonephritis, renal vasoconstriction, renal abnormalities) should be considered and differentiated from these diseases.

It is difficult to compare the hypertensive form of cranial nephritis with hypertension. In suran nephritis, urinary incontinence is less pronounced than in hypertension, and left ventricular hypertrophy is less common than hypertension.

Hypertensive crisis "crisis" is less common in chronic nephritis. In coronary nephritis, coronary atherosclerosis develops more slowly and less frequently than in hypertension. When comparing suranka nephritis with suran pyelonephritis, the number of erythrocytes and leukocytes in the urine sediment is taken into account. High levels of leukocytes, the presence of active leukocytes, changes in the structure of the renal vessels on X-ray examination of the kidneys indicate the presence of chronic pyelonephritis.

The nephrotic type of cranial nephritis should be compared with renal amyloidosis, diabetic glomeralosclerosis, liver cirrhosis.

In the nephrotic form of cranial nephritis, signs of renal inflammation (hematuria, hypertension), decreased renal glomerular filtration, and decreased renal concentration play a major role in the diagnosis. A biopsy of the kidney determines the diagnosis.

To differentiate from amyloidosis, the oral mucosa is examined histologically.

X-rays should be taken of the kidneys and urinary tract, X-rays of the kidneys with isotopes and contrast agents, ultrasound, and angiography of the kidneys.

Kidney Tissue Examination - The use of puncture biopsies can help diagnose the disease.

During the period of exacerbation of chronic nephritis, the patient should be hospitalized and treated, as in acute nephritis, should lie down and follow a strict diet. Symptoms of impaired renal nitrogen excretion (increased thirst, dry skin and mucous membranes, loss of appetite, nausea, vomiting, diarrhea, ammonia odor in the mouth, AD pressure is higher than normal. Daily protein intake is sharply limited (up to 25 g) due to the appearance of nausea, sleep disturbances, dizziness, increased residual nitrogen in the blood), 5% glucose by subcutaneous or intravenous drip for detoxification solution and 0.85% NaCL solution are infused, and the meda is repeatedly washed to remove nitrogenous wastes excreted through the intestinal tract. Drinking fluids are not limited, the daily salt intake is up to 8 g. Drinking alcohol is strictly prohibited.

Acute nephritis. - Infectious-allergic inflammation of the renal glomerulus (glomerulonephritis) is a disease that usually occurs after a person has a streptococcal infection (angina, scarlet fever, jaundice) and is exposed to cold. the disease begins with a sudden rise in temperature, swelling of the body, headache, hypertension, the appearance of simultaneous pain in the lumbar region. In the early days, oliguria is seen, and most of the urine that comes out due to the presence of a lot of hematuria is like a stream of meat. the patient should be hospitalized. The patient should lie down without getting up. In cases of significant swelling and hypertension, the patient is instructed to lie down without eating or drinking (starvation and thirst). It is important to keep in mind that eclampsia can sometimes lead to seizures in the early days of the disease due to brain swelling. Exacerbation of headache, changes in psyche, increase in blood pressure, dizziness of the eyes, the patient should beware of excessive impressions (bright light, loud noises, etc.).

Timely blood transfusions, intravenous or intramuscular injections of magnesium sulfate, and enemas of chloral hydrate prevent the onset of eclamptic coma.

7a - Instructions for prescribing a diet table. Acute glomerulonephritis (after days of sugar, potatoes), chronic nephritis with renal failure.

Purpose of appointment. Take care of the diseased kidney as much as possible, reduce the risk of hypertension and tumors.

General description of the table. A diet table prepared without salt, with limited protein, fats and carbohydrates limited to physiological norms. In this case, the food is prepared without salt. Special unsalted bread is covered, the amount of sodium in the food is about 400 mg, which corresponds to 1 g of table salt. The amount of liquid is up to 1000 ml (including liquid in the product). Patients with azotemia are given 1 to 3 g of table salt as directed by the treating physician. These patients are allowed to give the same amount of urine or 300-400ml of urine as the previous day. The diet is frequent, 5-6 times a day. Calories and composition of the table. Proteins - 20 g, fats - 70-80 g, carbohydrates - 350 g, calories - 2000 - 2200. Vitamins C and B are given in large quantities.

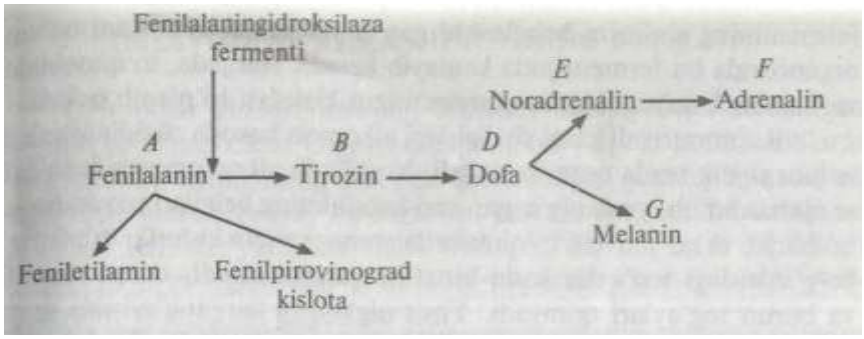
Method of preparation. All dishes are made from unsalted, shredded products. The bread is also covered without salt. It is not possible to fry dishes.

Supplied flavors. Low-protein, unsalted bread made with corn starch (100g per day) or 50g unsalted bread made from wheat flour with yeast per day. Allowed vegetable and fruit soups (without meat). Soups are boiled with onions, then lightly pasteurized and stuffed with sour cream and greens. Lean meat, chicken, rabbit, turkey, fish weighing 50-60 g per day are boiled or steamed and then steamed. 60g of milk per day (you can increase the amount of milk instead of meat). 2-3 eggs per egg (omelet or half-cooked). Low in protein pasta, sago, low in rice. These products are served in the form of porridge, pilaf or cutlets in a liquid with water and milk. A variety of dishes are prepared from potatoes and vegetables (fried pumpkin, fruit and vegetable

stew, steamed quince, surprise, steamed pumpkin, etc.) Various vegetable salads are prepared.

DISORDERS OF AMINOIC ACID METABOLISM

'Phenylketonuria. The disease was first diagnosed by Norwegian doctor F. Felling. He senses a certain odor in the urine of two mentally ill children. Biochemical examination of the urine of these children revealed the presence of large amounts of pyruvic acid. It is not found in the blood of healthy people. It is now known that the cause of this disease is related to the amino acid phenylalanine. Phenylalanine is the norm



Decomposition of phenylalanine.

phenylalanine is converted to tyrosine in the presence of the enzyme hydroxidase. In the presence of enzymes other than tyrosine, DOFA or 3,4-dihydro-phenylalanine, noradrenaline, adrenaline and melanin are formed. If the gene responsible for the formation of the enzyme phenylalanine hydroxidase is mutated, the enzyme will not be formed and the above-mentioned biochemical processes will not be observed. As a result, phenylalanine is converted into pyrovinogradic acid, which is excreted in the blood and excreted in the urine without being converted to tyrosine. This acid poisons nerve cells and leads to mental weakness. The absence of tyrosine, on the other hand, greatly reduces the amount of melanin. Due to the low content of melanin in the blood, the skin of such patients becomes yellowish-white. Patients experience mental retardation at an early age, and some experience seizures. Because the disease is not diagnosed and treated late, the patient's condition worsens. The disease occurs in autosomal recessive gentian influenza and occurs in children in a 1: 1000 ratio. Usually 1-2% of mentally retarded children have phenylketonuria.

In the diagnosis of this disease, mainly biochemical method is used. When a few drops of 5% FeCl3 solution are added to the urine, a green color appears. Diet plays a key role in the treatment of the disease, which means that the patient should not consume foods high in the amino acid phenylalanine. Preliminary biochemical analysis of urine and the advice of a geneticist play an important role in the prevention of the disease.

Alcoptonuria. In this disease, the metabolism of phenylalanine and tyrosine is disrupted. Tyrosine, which enters the body at the expense of phenylalanine and

food, is normally converted to P-hydroxy-phenylpyrovinogradic acid. This acid, in turn, is converted to homogentizinic acid by the enzyme homogentisotoxidase. In alcoptonuria, homogentisinoxidase
because the gene that determines the synthesis of the enzyme is mutated, this ferrnent in the body is greatly reduced. As a result, homogentisic acid accumulates in tissues and physiological fluids.
Because alcopton in the urine of a patient with alcoptonuria is oxidized in air, the urine quickly darkens. Alcoptonuria develops insignificantly in adolescence, the symptoms of the disease begin to appear in older adults, and homogentisic acid accumulates in the connective tissue, the joints in the joints turn yellow-purple, the ear canal and nose uncles darken. As you grow older, blood clots accumulate in the joints, leading to joint disease. This disease occurs in a 5: 1,000,000 ratio. Diet is the mainstay of treatment. low intake of foods high in phenylalanine and tyrosine! should do.
Albinism. The disease is caused by a mutation in a gene that controls the synthesis of the enzyme thyroatQQa, which converts tyrosine into melanin. In albinism, there is no color on the skin, hair, or cornea, and the eyesight is severely impaired. Albinism can occur in whole or in part. While partial albinism occurs in the presence of a recessive gene in the autosome, partial albinism occurs in the presence of a dominant gene in the autosome. Complete albinism occurs at a ratio of 1: 15,000, and partial albinism at a ratio of 1: 20,000. In complete albinism, sweat and hair turn white because the skin and hair are not pigmented, blood vessels are visible because there is no color in the iris, and therefore the eye is reddish. Partial albinism is characterized by white spots (areas without pigment) in some areas of the skin. In some cases, the hair is white and the skin and eyes are pigmented. Only the absence of pigment in the eye is observed. Such people are easily affected by light. In some cases, deafness in albinism and the development of certain organs have a number of shortcomings. There is a village in the United States where people with complete albinism live. They only survive at night. and during the day they do not go out of their houses. U.S. scientists are examining them. Nowadays, they have created a drug that is applied to the skin of utfiun. Once this drug is applied to the skin, a person can walk in the sun for 2 hours.
Albinism is multiple genocopy, i.e., the same phenotype in patients is produced by different genotypes. Although the disease is considered autosomal recessive, it can also occur in the autosomal dominant state. This suggests that albinism is caused by mutations in non-allelic genes that exhibit the same clinical features. 224

DISORDERS OF CARBON METABOLISM

There are a variety of diseases that occur with impaired carbohydrate metabolism. Mutations in a gene involved in the synthesis of an enzyme that breaks down mono-, di-, and polysaccharides in the body result in galactosemia, fructosuria, pentosuria, diabetes, and other diseases.

Accumulation of glycogen leads to glycogenesis and accumulation of amino carbohydrates leads to mucopoly-saccharidosis.

Galactosemia. This disease is caused by a deficiency of the enzyme galactose-1-phosphaturidyltransferase, which breaks down galactose-1-phosphate in the body. The body cannot absorb galactose. therefore, it accumulates in the blood and tissues and begins to have a toxic effect on the body. As a result, changes occur in the liver; Protein is excreted in the urine due to impaired renal function. Therefore, in this disease, the urine is rich in protein and amino acids. The disease appears in the first days of breastfeeding. Symptoms include: vomiting, jaundice, weight loss, diarrhea, dehydration, visceral dysfunction, mental retardation, severe form of the disease, the child dies in a few months, the disease occurs in a ratio of I: 70,000 .

Diagnosis is based on the amount of galactose in the blood. In galactosemia, the amount of galactose increases. The disease can be diagnosed in time and treated with diet.

Mucopolysaccharidoses. This is due to the disruption of mucopolysaccharide metabolism. Mucopolysaccharides accumulate in large numbers in lysosomes because the lysosome does not have a fermenter that breaks them down. Patients with mucopolysaccharidosis develop changes in the structure of the skeleton, skull, face, eyes, and internal organs, and mental retardation. Mucopolysaccharides accumulate in the spleen, spleen, bone marrow, and connective tissue and are excreted in blood and urine. Children with the disease can live up to 12 years. There are currently 7 types of mucopolysaccharidosis, all of which have the same phenotype. All types of mucopoly-saccharidoses are inherited through an autosomal recessive gene.

DISORDERS OF PURIN AND PIRIMIDIN METABOLISM

Hereditary disease caused by impaired purine and pyrimidine metabolism is caused by a deficiency of the hypoxanthine-phosphoribosyl-transferase (GFRT) B enzyme in the body. this enzyme accelerates the conversion of guanine and hypoxanthine, the purine compounds in free hoIat-B, to nucleotide-M. When this enzyme is deficient, the amount of uric acid in the body increases. In a healthy person, the norm for 1 g of uric acid is 20-3p g tcng in diseases caused by disorders of purine and pyrimidine metabolism. Symptoms begin in infancy and are manifested by increased muscle contraction and increased sensitivity. J Hemoglobinopathy. The disease is caused by changes in the structure of hemoglobin. For example, in the case of anemia with erythrocyte-shaped erythrocytes, the exchange of valine I with glutamic acid in the 6th position of the B-bond leads to poor solubility I in hemogiobin and high polymerization properties. Heterozygous organisms are usually healthy. But as the amount of oxygen in the air decreases, the symptoms of the disease begin to appear. In homozygous organisms, the symptoms begin very

early and are followed by chronic oxygen deprivation, anemia, circulatory disorders, and thrombosis. The above-mentioned changes in hemoglobin are common in areas where malaria is common, as this change in hemoglobin makes the erythrocyte resistant to the parasite that infects them and increases the viability of heterozygous organisms. in a number of regions of Central Asia and the Caucasus, heterozygous (carrier) and diseased homozygous individuals are more common. The disease is inherited in an autosomal recessive state.

Hemophilia. This disease occurs as a result of disruption of the structure of the enzyme that synthesizes the protein that promotes blood clotting. It has long been known that hemophilia has a hereditary nature. According to some sources, the disease was only known in the 5th century. Several types of hemophilia are now known (A, B, C, D). A baby born with hemophilia can die if the umbilical cord is not cut. Bleeding can also occur in slightly injured internal organs.

Hemophilia is more common in men because the recessive mutant gene that causes the disease is located on the X chromosome and not on the Y chromosome. A father with hemophilia gives his daughter an X chromosome, a mutant gene that causes hemophilia. However, a girl with this X chromosome does not suffer from hemophilia. because it is the dominant allele (XH) of that gene, and in a heterozygous organism (XHX) the dominant gene trait appears. But even though she is not sick, she is still a carrier. The mother carries the hemophilia gene to her sons. Hemophilia occurs in a population in an I: 5,000 ratio.

It is denoted by 1 Latin letter and their numbers are given (A ,, A ,. A ,, A4). Examples of polygenic heredity include skin color. Skin color is controlled by five pairs of dominant genes that complement each other (A, A ,; B, B ,; C, C ,; D ^ D ,; E, E ^). The formation of melanin in the skin is caused by a pair of genes, but the remaining 4 pairs of genes determine how much melanin is produced.

The color of the skin depends on the amount of pigment formed in it. Europeans do not have dominant genes, but have 4 pairs of recessive genes. Height, weight, lifespan, blood pressure, finger length, and so on are all signs. ^

Criteria for assessing the knowledge and skills of groups.

86 – 100 score	The student is able to think independently about the etiology, clinic, diagnosis and treatment of gastrointestinal ulcers, rheumatoid arthritis, nephritis, diabetes mellitus, schizophrenia, and to independently diagnose and carry out preventive measures.

71 – 85 score	The student is able to think independently about the etiology, clinic, diagnosis and treatment of gastrointestinal ulcers, rheumatoid arthritis, nephritis, diabetes mellitus, schizophrenia, and to independently diagnose and carry out preventive measures.
55 – 70 score	Moderately answers questions on the topic, does not fully understand the etiology, clinic, diagnosis and treatment of gastrointestinal ulcers, rheumatoid arthritis, nephritis, diabetes, schizophrenia. can not take preventive measures.
0 – 55 score	Has a very shallow knowledge of gastrointestinal ulcers. Cannot take preventive measures.

PRACTICAL LESSON 6

Prenatal diagnosis of hereditary diseases.

Class time - 2 hours	Number of students: 18
Class type	Practical training
Plan	Medical - genetic counseling. Modern methods of prenatal diagnosis (UTT, chorionic biopsy, fetoscopy, amniocentesis). Guidelines for prenatal diagnosis, possible errors, complications, screening diagnostic methods. Guidelines for medical and genetic counseling, methods of calculating the risk of birth of a sick child, methods of identifying carriers of pathological genes, family planning. The Teaching Method
The purpose of the topic: - Know how to develop risk factors, - timely diagnosis, - Methods of testing and identification of risk factors in young families - To teach to use easy and convenient methods of detection of infertility in couples.	
Task of the topic - - Formation of interest in the profession, humanity, a sense of responsibility; -Education of interest in expanding their knowledge, development of students' thinking skills, logical thinking; - Guide students to a creative approach to the study of the topic.	

Teaching methods	Demonstration, lecture, conversation
Teaching forms	Mass, collective
Teaching aids	computer, multimedia, slides, subject patients, etc.
Teaching environment	Methodologically equipped auditorium.
Monitoring and evaluation	Oral control: questions and answers

Stages and timing of work	**Educator**	**Learners**
Preparation stage	1. Controls the cleanliness of the audience. 2. Checks students' readiness for classes 3. Controls attendance.	
1. Introductory phase (10 minutes)	1. Prepare curriculum on the topic. 2. Prepare presentation slides for the introductory presentation. 3. Develop a list of references used in the study of science.	
2. The main stage (55 minutes)	1. Divide students into small groups and ask questions on the topic. 2. Demonstration posters are used. 3. Slides, multimedia are used 4. Conducts treatment 5. Summarizes the information provided on the basis of the topics, encourages the active participant students and gives an overall assessment.	They are divided into small groups They watch They participate They listen and answer questions
3. The final stage (10 minutes)	1. Concludes. 2. Provides independent work. 3. Gives homework.	They listen Take notes Take notes

Questions on the topic:

1. What is the main function of medical-genetic counseling?
2. Stages of implementation of medical-genetic counseling.
3. The importance of prenatal diagnosis.
4. Principles of treatment of hereditary diseases.

Interactive methods on the topic:

"Wheel"

Required for the game:

A set of questions and a situational question are presented on a separate paper
It should be individual for each student and group
Clean paper and pen
The game is on.
Total game time is 45 minutes
The group is divided into 3 small groups (4 students in each small group).
Each small group sits at a separate table. He makes a blank piece of paper and takes a pen.
The date, group number, name of the game and the name of the group will be written on the paper.

One of the participants takes a question or situational question from the envelope (a question or situational question for the group is chosen by the teacher)
A separate question or situational question is selected for each group, but their complexity should be the same
Discuss with the group and 1 student writes the answer on a piece of paper.
The analysis takes 15 min
Students' participation in the game will be checked by an assistant
Over time, students hand over their work to teachers
All results are analyzed, the most correct answer is selected and the maximum score is given.
The analysis takes 15 min
Students' scores affect the current grade in the practical class
1) Place 86-100 points. 2) Place 71-85.9 points. 3) place 55-70.9 points
At the end of the answers, the assistant scores and signs
the scores obtained by students are relevant to the current grade in the internship.

1. What is the main function of medical-genetic counseling?
2. Stages of implementation of medical-genetic counseling.
3. The importance of prenatal diagnosis.
4. Principles of treatment of hereditary diseases.
5. Provide perspective advice
6. Retrospective counseling
7. The main function of medical genetics counseling
8. Giving referrals to clients
9. Four stages of medical genetics consultation
10. Empirical method
11. Genealogical method
12. Perspective advice is a must
13. Must give retrospective advice
14. Must know the main function of medical genetics counseling
15. Must be able to refer to clients
16. Medical genetics consultation must know the four stages
17. Must know the empirical method
18. Must know the genealogical method
19. Must be able to reset the Proban card
20. Must be able to detect and analyze sex chromatin
21. Must be able to interpret the biochemical test and its results
22. Dermatoglyphic examination and interpretation of its results

Course content:

Medical-genetic counseling is a specialized medical care provided by a geneticist to a patient with a hereditary pathology and his family, which is carried out in a special medical institution - medical-genetic counseling. The main task of medical-genetic counseling is to determine the prognosis (probability of occurrence of

hereditary pathology) in a family that is restless with a hereditary disease, and to implement preventive measures based on this prognosis. Preventive measures are the prevention of the birth of a child with a hereditary disease in the family. Visitors are also explained the content of genetic risk and whether or not they can have children. The task of the geneticist is to carry out a special genetic examiner to isolate the risk group to diagnose a hereditary disease.

a) if a child with a genetic pathology was born, died or is alive in the family, or if the birth is suspected;

b) when several members of the family have similar pathological symptoms or diseases, inability to tolerate certain foods or medications;

c) retardation of physical and mental development in children, when there are various key developmental disorders;

g) in cases of premature termination of pregnancy, primary amenorrhea, genital hypoplasia or primary infertility in the family;

d) when the husband and wife are blood relatives. Reducing the number of people with inherited diseases today depends on how medical and genetic counseling is organized. Genetic counseling is one of the special medical services provided to the population. The first genetic counseling clinics were established in 1967.

The main tasks of medical-genetic counseling are to know the causes of hereditary diseases in the newborn and to prevent the development of hereditary diseases, etc. enters.

Counseling for parents by a doctor is divided into several stages.

In the first stage of the consultation, the diagnosis of the disease is made by examining whether the disease is hereditary or not. At this stage, a very thorough study of the genetic structure of the family with the physician-genetic disease determines whether the disease being studied is dominant, recessive, or gender-dependent.

In the second phase of counseling, the probability of having a sick child in the family being studied and whether the disease is monogenic or polygenic is determined.

In the third stage of the consultation, the doctor provides written information about the next generation.

In the final stage, the doctor should explain to the parents very carefully about the disease that may occur in their children.

Medical and genetic counseling should be done as often as possible. During this time, the doctor will be able to explain the genetic risk, and the parent will have a clear idea. The decision on whether or not to have a final child should be made by the parents themselves, but in some cases, if the father or mother is ill, the doctor may recommend that they have a child.

1. When one of the parents has an autosomal dominant disease in which a biochemical disorder has been identified;
2. When the mother is over 35 years old;
3. When the parents are close relatives;
4. When it is determined that the father or mother is exposed to mutagens and teratogens (even during the mother's pregnancy);

5. When there are cases of miscarriage; Determining the sex of the fetus in the mother's womb is of great importance in resolving the issue of childbirth in sex-linked hereditary diseases.

X-chromatin can be detected in cells in amniotic fluid obtained by amniocentesis (this method is performed at 18-20 weeks). The method is not very complicated, but it has some shortcomings. Because Barr's body is found not only in healthy girls, but also in boys with Klinefelter's syndrome. Barr corpuscles are found in both healthy boys and girls with Shereshsky-Turner syndrome. X chromosomal aberrations and mosaicism can also be mistaken. Detection of Y-chromatin is currently underway. This method is used when there is a suspicion that the number of Y-chromosomes has increased. When stained with acrixin, the long shoulder of the Y chromosome is strongly fluorescence at the interphase. Depending on the number of fluorescent Y-cells, the number of Y chromosomes in the set is determined: 46, one body when XY is stained; In 47 XYY - two, in 48 XYYY - three bodies are defined. The disadvantage of this method is that it does not detect the non-fluorescent part of the Y chromosome, but contains genes that determine the development of sperm.

Since most methods of prenatal diagnosis use amniotic fluid cells, we are familiar with the method of amniocentesis. This method is performed by an obstetrician in a specialized obstetrics department. If no precautions are taken, the fetus may die as a result of satellite damage.

Once the gestational age has been determined, an ultrasound scan is performed to determine the location of the placenta and a needle is inserted into the amniotic fluid through the abdomen and uterine wall and fetal membranes with a special mandrel needle. Then 10-20 ml of amniotic fluid is drawn using a syringe by pulling out the mandrel.

Usually (in 95-97% of cases) it falls into the amniotic cavity on the first attempt. If no complications are felt, the pregnant woman can be answered at home after 1 to 2 hours.

K.N -sensitization can sometimes be observed as a complication if the sperm is removed after amniocentesis.

If amniocentesis is performed after 32 weeks of gestation, fetal injury may be observed.

Currently, the reduction in the number of people suffering from hereditary diseases depends on how medical and genetic counseling is organized. Genetic counseling is one of the special medical services provided to the population. The first genetic counseling clinics were established in 1967. Similar receptions have been opened in many provinces.

The main tasks of medical-genetic counseling are to determine whether a child is born with a hereditary disease, to know the causes of hereditary diseases and to prevent the development of hereditary diseases, and so on. Usually, parents with a child who is seriously ill or physically weak in the family need medical and genetic counseling and are concerned about how their next child will be born. Even a person with a serious inherited disease in his offspring is interested in what the next generation will look like. In solving such difficult problems, the doctor has a

great responsibility. Because the doctor tells the parents who came to see him, whether their next child is healthy or not! must give a clear answer about being born. The doctor may be mistaken in this regard, otherwise the parents and the sick child at birth may suffer for the rest of their lives. Therefore, the information about the heredity of this disease - when enough, the doctor explains to the parents that it is dangerous for them to have children.

In some cases, healthy parents are afraid of having a sick child in the family because their offspring have an inherited disease. In such cases, it is necessary to study the disease in detail and tell such families that they are lucky to have children. However, the probability of having a sick child in the family is 25%, and even if it is 25%, the doctor should not recommend the expectant parent to have a child. Although this figure may seem small to parents, it is important to keep in mind that the first child in the family may be born ill. Therefore, the geneticist should explain the disease to the parents in detail and conduct the consultation in several stages.

The first step in counseling is to determine whether the disease is hereditary or not, and to make a definitive diagnosis. At this stage, the doctor carefully examines the genetic structure of the family with the genetic disease to determine whether the disease being studied is dominant, recessive, or gender-dependent.

In the second phase of counseling, the probability of having a sick child in the family being studied and whether the disease is monogenic or polygenic is determined. Because the disease occurs with a dominant gcn (A), the AA and Aa genotypes are diseased and aa is considered healthy. If one parent in the family is heterozygous (Aa) and the other is homozygous (aa) healthy, the probability of having a sick child is 1: 1. If the disease is caused by a recessive gene (a), both healthy parents can give birth to a sick child. If the parents are heterozygous, a sick child can be born from a healthy parent (Aa, Aa). From each heterozygous parent, two (A, a) 4 gametes are formed. These gametes combine to form 4 different genotypes: AA, Aa, Aa, aa. These organisms are considered healthy because 3 of these genotypes contain the dominant (A) gene. In an organism with two recessive genes, the disease occurs. This means that there is a 25% chance that a healthy child will be born to a healthy parent. If one parent is gotnozygotic on the dominant gene, all children will be healthy.

The disease is caused by more than one gene. it is very difficult to say whether the character studied in subsequent generations will emerge as dominant or recessive. Because it is impossible to predict the genotype of the parents and how the trait under study will be different from the next generation. Therefore, it is possible to determine the likelihood of the disease in future generations only on the basis of data obtained over several years on such diseases. However, for some common polygenic diseases (epilepsy, schizophrenia), it is possible to make a definite point about the likelihood that sick children will die in the next generations. In general, if a healthy child is born in the family, the probability of the next child to be born sick is 1%, because in the population 230 cases of this disease are 1%. If one of the parents is sick, The probability of the first child to be born sick is 19%.

the probability is 59%. In the third stage of counseling, the doctor provides written information about the next generation. If the probability of developing the disease in the unborn child is 5%, the genetic risk is low, the risk is increased to 10%, but in mild form, the risk is high to 20%. With a 10% risk, a doctor can allow a parent to have a different color. But she is pregnant I her unborn child will have to undergo a genetic test. In the final stage, the doctor should explain to the parents very carefully about the possible disease that may occur in their children. Because all parents want to have children. Therefore, the doctor also said that 4-5% of children born in the population are born with inherited diseases, and the birth of sick children is not only in the families of those who come to the doctor. should be reminded that it is also present in other families.

Criteria for assessing the knowledge and skills of groups.

86 – 100 score	The student knows the principles of medical genetics counseling, is able to conduct independent observation. Knows the guidelines and contraindications for conducting medical genetics counseling. Can plan a family and use medical genetics.
71 – 85 score	The student knows the principles of medical genetics counseling, is able to conduct independent observation. Knows the guidelines and contraindications for conducting medical genetics counseling. Can plan a family and use medical genetics.
55 – 70 score	Intermediate answers to questions on the topic, not fully aware of the principles of medical genetics advice. Does not know the guidelines and contraindications for conducting medical genetics counseling.
0 – 55 score	The student has a very shallow knowledge of the principles of medical genetics counseling. Does not know the guidelines and contraindications for conducting medical genetics counseling.

A healthy child is a source of joy and happiness for parents, as well as a source of strength for the family. No matter how unpleasant it may seem, the problems and worries that result from the birth of an unhealthy, defective child can disrupt the peace and tranquility of the family, and the situation can even lead to divorces. In our society, there are screening centers to prevent such cases - to monitor the birth of healthy children in the family. Through our website, we will get acquainted with

the activities of such centers and provide relevant information about screening tests.

Phenylketanury is a congenital metabolic disorder that causes mental retardation and is passed down from generation to generation. Symptoms usually appear between the ages of 2 and 6 months.

Hypothyroidism is a disease characterized by insufficient production of thyroid hormones. Failure to diagnose and treat it in a timely manner can result in severe physical impairment and mental retardation.

The word "screening" (eng.t) means "screening". Screening is a mass screening of the population for early and rapid detection of the disease using special methods.

With modern medical equipment and qualified staff, the Screening Centers work in three main areas:

1. Prevention of fetal malformations in the prenatal period;
2. Mass screening of newborns for congenital hypothyroidism and phenylketonuria;
3. Medical and genetic counseling for people born with hereditary and congenital diseases, as well as for families with such children

How many different screening tests are available?

There are two main types of screening tests: neonatal, prenatal, and screening.

Neonatal diagnosis is a specific type of mass medical examination of newborns that can be used to identify the most common inherited and congenital diseases. Early detection and treatment of various diseases that can lead to adverse outcomes due to neonatal screening can prevent the development of a number of dangerous diseases that can lead to disability.

Blood should be taken 3 to 4 days of life for premature babies and 7 to 10 days of life for premature babies. If the blood is taken earlier than indicated, the test may be incorrect.

For information: Patients identified during neonatal screening (including children with viral hepatitis and phenylketonuria) are on the dispensary list at screening centers. They will be provided with medicines and free medical food.

When and how is a neonatal diagnosis made?

A few drops of blood are taken from the newborn and dipped in a special paper. This paper is then sent to the screening center. Blood tests are performed here on special modern equipment. If changes in the child's blood test suggest a disease, the mother and child are called to the clinic or screening center for re-examination. Only a re-analysis of the blood, a specialist examination will make it clear that the baby is really sick and needs treatment.

Why do you need a prenatal checkup?

Prenatal screening can analyze the blood of a pregnant woman to determine if there are any deep developmental defects in the fetus. The main purpose of prenatal (prenatal) diagnosis is to make sure that the developing baby is healthy, to prevent the birth of a child with congenital malformations and diseases that lead to disability. In pregnant women, prenatal examinations are performed three times - at 10, 14, 16, 20 and 28 32 weeks of fetal development.

The following pregnancies should be screened before delivery

• If the child was born with a hereditary disease or congenital malformation.
• Pregnancy with miscarriage or stillbirth.
• If one of the parents is diagnosed with a hereditary disease.
• Relatives.
• A woman is under the age of 18 and over the age of 35.
• Occupational exposure (radiation).
• When using drugs or chemicals that affect the fetus.
• Infectious diseases of the fetus - measles, toxoplasmosis, chlamydia, cytomegalovirus.
• If a man or woman is found to be using drugs or alcohol.
• Endocrine disorders of the fetus - thyroid gland and diabetes.
• Pregnant women at risk of having children with congenital heart disease and chromosomal syndrome (Down syndrome, etc.);
• In-law marriage (in cases of genetic abnormalities in the family!);

What is the procedure for screening for fetal malformations and hereditary diseases?

Initially, a pregnant woman must be registered at the polyclinic at her place of residence until the 14th week of pregnancy. If the doctor identifies a factor that affects the health of the fetus, the pregnant woman will be referred for a stage II examination at the screening center. Screening centers perform ultrasound and biochemical screening tests at 16 to 20 weeks of gestation. It detects the levels of specific biochemical markers (alpha-fetoprotein (AFP)) and chorionic gonadotropin (XG) in a pregnant woman's blood.

Medical-genetic advice…

The third area of activity of the center is MEDICAL-GENETIC ADVICE, which involves checking parents with pathological relatives for congenital diseases. It is possible to accurately diagnose suspected hereditary diseases, determine how the disease is transmitted from generation to generation and the risk of its recurrence in the family. If the doctor-geneticist concludes that there is a hereditary disease in the family, he will be able to calculate the probability of the disease in future children and give the necessary advice.

Let me know!

According to doctors of the center, the factors that lead to birth defects and hereditary diseases are the marriage of girls under the age of puberty, untimely gynecological examination of women of childbearing age, childbirth of women with extragenital diseases. One of the main reasons is in-law marriage. Because every child gets half of their genes from their mother and half from their father. Children of related parents are more likely to have "disease" genes. The birth of a baby with Down syndrome is mainly related to the age of the mother. It is more common in children of women over the age of 35. It is not possible to identify all

the genes in humans. About 3,500 genetic defects have been identified so far, most of them in childhood.
Fact: There are now more than 3,500 known inherited and congenital diseases, some of which are diagnosed in childhood. Some inherited diseases are passed on from generation to generation, that is, from parents to children, and some diseases are also found in the children of physically healthy parents (when the disease is secretly inherited).

№	Self-study topic	Hour
1	Albinism, etiopathogenesis, clinic, diagnosis, treatment and prevention.	**3**
2	Hereditary diseases of the skin (dermatoses), Diabetes mellitus.	**3**
3	Hereditary diseases of the nervous system (myopathies), new modern research methods used in medical genetics	**4**
4	Familial hypercholesterolemia, dysmetabolic nephropathy	**4**
5	Hereditary diseases of the blood. Multifactorial diseases	**3**
6	Metabolic features in patients with hereditary diseases, features of the nervous system in patients with hereditary diseases.	**3**
	Total	**20soat**

Independent work on science is carried out in the classroom and outside the classroom.
• The following forms are used in the organization of independent work of students:
• Reflecting the practical skills approved in the laboratory of the clinical base in the notebooks of quantitative and qualitative marketing and practical skills under the supervision of the teacher;
• Prophylactic examination of the population in primary health care facilities, dispensaries, participation in patronage;
• Conducting public health interviews and lectures;
• independent study of selected theoretical topics with the help of textbooks;

• preparation of information (abstract) on a given topic;
• Work and report on special or scientific literature (monographs, articles) on sections or topics of science; Preparation of scientific articles, abstracts for the conference;
• Solve situational problems focused on situational and clinical problems;
• Solve case (case-study based on real clinical situations and clinical situation issues).
• The student also has an independent work:
• Development and completion of graphic management;
• Create and solve crossword puzzles;
• Preparation of presentations and videos and their widespread use in independent work, etc.
• Participation of students in competitions, exhibitions, conferences and other events in the active study of science.

G L O S S A R Y

Autopolyploidy is a form of polyploidy that results from the reproduction of the genome of the same species

An ancestral effect is the proliferation of a rare gene in an initially small population.

Allyel is one of the states of the gene. Allelic genes are located at ~ identical loci of homologous chromosomes and exhibit alternative traits.

Alloploidy is the multiplication of a haploid set of chromosomes as a result of the merging of the genomes of different species or generations. Such hybrids usually do not leave nacl due to meiotic disorders.

Amniocentesis is the treatment of amniotic fluid in the amniotic sac of the uterus for prenatal diagnosis. .

An antigen is a substance that is genetically foreign to the same organism. Due to their chemical nature, they can be protein, glycoprotein or polysaccharide. Viruses, microorganisms, and even the body's own cells can be antigens.

Anyeuploidy (Goetheroploidy) is the presence of an unbalanced set of chromosomes in cells.

Achondroplasia is an autosomal dominant disease characterized by decreased growth of long, tubular bones. In the disease, even if the size of the head and body are normal, the gray legs are very short.

Galactosemia is an autosomal recessive disease caused by a disruption of the synthesis of an enzyme that controls the secretion of lactose.

Genetic Expression is the realization of information encoded in DNA in the processes of protein biosynthesis, transcription and translation.
Genetic code is a system of writing genetic information as a sequence of nucleotides in the DNA (RNA in retro-viruses) chain. The code is in the triplet state, with information about a single amino acid being identified by three nucleotides.
A genome is a collection of (l) chromosomes in a haploid set. The genome characterizes the species, and the genotype - in-dividium. In diploid organisms, the genome represents the chromosomes of gametes and is used as a unit of genetic analysis.
Genotype is the genetic constitution of an organism, the sum of all alleles in its diploid set.
Hemoglobinopathies are a group of different diseases caused by gene mutations that control hemoglobin synthesis.
Homologous Chromosomes are pairs of chromosomes that are the same size, shape, and genes.
A homozygote is an organism (aa or AA) that has the same alleles of the same gene.
Discordantness is the difference in signs with a narrow reaction rate in zigzags.
Duplication is a type of chromosome aberration in which one part of a chromosome doubles.
Gender is a set of traits that allow an organism to participate in sexual reproduction and pass genetic information on to offspring through gametes.
Inversion is one of the aberrations within a chromosome in which a part of a chromosome (or gene) is broken, rotated 180 °, and repositioned.
Heredity is a trait that ensures the material and functional continuity of organisms during the process of generation through the process of inheritance.
A karyotype is a set of traits (number, shape, size) of a set of chromosomes that is unique to each species.
Crossingovyer - 1 exchange of parts of homologous chromosomes in the prophase of meiosis (resulting in new combinations of genes and traits).
Meiosis is a method of maturation of gametes, a two-fold decrease in the number of chromosomes as a result of the division of an immature gamete.
Mitosis is an unstable division, a process specific to eukaryotes. Due to mitosis, virgin cells receive equal and identical genetic information, and each cell has a 2p set.
Monosomy is the absence of one of 2 homologous chromosomes in a diploid set of chromosomes (2i-1, e.g., 45, XO).
A mutation is a change in genetic information at the gene, chromosome, or genome level that causes a new trait or trait to emerge.
Pleiotropy is the ability of a single gene to affect a polynomial. A classic example of this is Marfan syndrome. Pleiotropy can be primary or secondary.

A proband is a person (patient) who collects data for genealogical analysis.
Transcription is the transfer of information from DNA to RNA. RNA synthesis from a DNA matrix.
Translocation is one of chromosome aberrations, a change in the location of genes.
Translation is the transfer of information from RNA to a polypeptide, the synthesis of a polypeptide in the ribosome on the basis of the RNA matrix, the "translation" of the language of nucleotides into the language of amino acids.
Transposition is the replacement of a gene on a single chromosome.
Trisomy is a genome mutation in which one or more chromosomes in a diploid set become 3 instead of 2.
A phenotype is a set of all the features and characteristics of an organism that arise as a result of the interaction of a genotype with the environment.
Chromatin is a dye in the interphase nucleus, a dispersed state of deoxyribonucleoprotein.
Expressiveness is the degree of phenotypic occurrence of a character, incessant expressiveness reduces the level of anxiety and the occurrence of the disease in the offspring.
Epistasis is a form of interaction of noallel genes in which one allele (epistatic) suppresses the action of another nonallel (hypostatic) gene.

Modern information and pedagogical technologies in science teaching

The use of advanced and modern teaching methods, the introduction of new information and pedagogical technologies are important for students to master the science of medical genetics. Textbooks, teaching aids, lecture notes, handouts, computer programs, electronic materials, as well as modern light microscopy and video systems are used in the study of science. Advanced pedagogical technologies are used in lectures and practical classes.

Keys method (Case study)

The main purpose of the case study is to study the content of the topic in depth, to have a thorough knowledge of the standards of diagnosis and treatment, the ability to think independently, to study the problem in depth, to show and analyze the shortcomings of the doctor , the development of measures to prevent them. To successfully complete this case study, students must first have the following knowledge and skills:

The student should know: the etiology of viral hepatitis, clinic, patient management and prevention based on the accepted standard of diagnosis and treatment.

The student must: study the topic independently; identifies the nature of the problem; learns to make independent decisions by critically reviewing data; has its own point of view and draws logical conclusions; works independently with educational data; compares, analyzes and summarizes data;

Must have: communication skills; presentation
Skills; collaborative skills; problematic situations
analytical skills.

Technological scheme of case-based training:

1. Introduction to the case stage
2. Introduction to the case study
3. Organization of work with the case study (distribution of assignments, instructions).
4. Solve the case stage in a modern way (the group is divided into subgroups) (discussion - its structure is determined by the questions at the end of the case - analysis and solution of the problem situation, aimed at developing recommendations for action in such a practical situation).
5. Teacher's resume.
6. Assessment of student achievement

"Round table"

Needed for the game.

1. A set of questions and a situational question are presented on a separate paper

2. It should be separate for each student and group

3. Clean paper and pen

The game is on.

1. The group is divided into 3 small groups (4 students in each small group).

2. Each small group sits at a separate table. He makes a blank piece of paper and takes a pen.

3. The date, group number, name of the game and the names of the participants of the group are written on the paper.

4. One of the participants takes a question or situational question from the envelope (the teacher chooses the question or situational question for the group).

5. A separate question or situational question is selected for each group, but their complexity should be the same

6. White paper is distributed to everyone in the circle.

7. Each student writes their answer on this piece of paper.

8. Students are given 3 minutes to write the answer to each question

9. Over time, students hand over their work to teachers

10. All results are analyzed, the most correct answer is selected and the maximum score is given.

11. The analysis takes 15 minutes

12. All participants will receive the following points. Maximum score 0.8 points

1) 0.8-0.7 - “5”

2) 0.6-0.4 - “4”

3) 0.4-0.1 - “3”

4) 0 - “2”

13. The score obtained by students affects the current grade in the practical training

"Pen in the middle of the table."

Needed for work.

1. Questions

2. Clean paper pen

3. Workbook

The game is on

1. Total time 45 min

2. All students are divided into 3 small groups of 4 students each

3. Each small group sits at a separate table. Makes a blank piece of paper and gets a one-color pen.

4. The date, group number, name of the game and the names of the participants of the group are written on the paper.

5. Assignment: All small groups answer one specific question

6. Each student writes his / her last name, answer option on a piece of paper and gives the paper to a neighbor. He then moves his pen in the middle of the table.

7. The educator supervises the work of the group and each participant in the group. The general correct answer is written in the notebook.

1st place 5.

2nd place 4.

3rd place 3

8. The assistant scores and signs the paper.

9. The score obtained by students affects the current grade in the practical training

“Who’s more? Who's smart? ”

Required for the game:

1. Cards with questions on the topic (The number of cards should correspond to the number of students in the group, 5 questions on each card)

2. stopwatch.

Progress:

1. Total game time is 45 minutes

2. The game is played orally

3. Students take turns drawing question cards. For 3 minutes, each student answers 5 questions orally

4. The teacher counts the number of correct answers

5. All students participate in the game

6. Questions that are not answered correctly are analyzed

7. Students' answers are evaluated in the following form:

1) 5 correct answers 0.8 points

2) 4 correct answers 0.7 points,

3) 3 correct answers 0.5 points,

4) 2 correct answers 0.3 points,

5) 1 correct answer 0.1 points

Find Your Mate

Take different shapes (pen, tree, flower, heart, apple, etc.), colored paper and divide by 2. Part 1 contains a question, part 2 contains an answer. The number of sections should be the same as the number of students. The questions on the piece of paper are read one after the other to each student. The student raises his hand to answer any of the questions. When answering a question, if the color of the pieces of colored paper match, the answer is correct.

The "I teach, I learn" method

Required for the game:

1. A set of questions written on separate papers. (4 pieces)

2. Pure white paper posters (4 pieces)

3. Group name papers

4. Total game time - 45 minutes

5. Group participants choose group names and are divided into 5 subgroups (2 students for each subgroup).

6. Each small group sits at a table opposite each other. She makes plain paper posters and waffles.

7. The name of the group is written on the paper.

8. Participants select questions and each (teacher) group explains, discusses and prepares a presentation to the opposite (learner) group.

9. You will be given 5 minutes to prepare a discussion and presentation

10. The question is discussed among the small groups, and the study group writes their opinion on the poster.

11. When the time is right, the groups play. (5 min)

12. Presentation of small groups prepared for the presentation.

13. Answers are encouraged with stars.

14. The 5th subgroup is appointed as an expert.

15. The stars in the groups are counted and evaluated. The students with the highest number of stars will be evaluated, and the other groups will be evaluated better and more satisfactorily according to the number of stars.

16. The score obtained by students affects the current grade in the practical training

17. Student work is maintained by the teacher.

III. Number of study hours

Hour	Distribution of the amount of workload by classroom hours (hours)			
	Lecture	Practical lessons	Clinic training	Self -study
56	6	12	18	20

Basic theoretical part (lectures)

Thematic plan of lectures.

№	Topic	Hour
1	History of the development of genetics. Classification of hereditary diseases, etiopathogenesis. Chromosomal diseases (general characteristics). Screening program	3
2	Monogenic diseases. Criteria, hereditary type of character. Criteria and nature of multifactorial diseases.	3
Total:		**6 hours**

Content of lectures

Topic 1: History of the development of genetics. Classification of hereditary diseases, etiopathogenesis, mutation process, clinic. Chromosome diseases. Skinning software. Frequency and prevention of congenital and hereditary pathologies. Autosomal and sex chromosomal diseases, diagnostic criteria and methods of treatment.

Topic 2: Monogenic diseases, criteria, character, hereditary type. Criteria and nature of multifactorial diseases. FKU, alkaptonuria, galactosemia etiopathogenesis, clinic, diagnosis and treatment methods. Medical genetics advice. Methods of prenatal diagnosis. Hereditary diseases.

Thematic plans for practical and clinical training

№	Topic content	Hour	Clinical training	Total

1.	Methods of diagnosis of hereditary diseases.	2	*3*	*5*
2	Chromosome diseases.	2	*3*	*5*
3.	Monogenic disease. etiopathogenesis, clinical, diagnostic methods. Disorders of protein metabolism, disorders of carbon-water metabolism Disorders of fat metabolism.	2	*3*	*5*
4	Diseases of connective tissue metabolism .Mukopolysaccharidoses.	2	*3*	*5*
5	Multifactorial diseases	2	*3*	*5*
6	Prenatal diagnosis of hereditary diseases. Medical - genetic counseling. risk - anxiety	2	*3*	*5*
	Total	**12 hours**	**18 hours**	**30 hours**

Final assesment questions

1. Hereditary features of traits in humans.

2. To study the inheritance of traits in twins.

3. To study the method of determination of sex chromatin.

4. Study of human chromosomes, acquaintance with the method of preparation of drugs.

5. Create a family tree.

6. Etiology and pathogenesis of chromosomal diseases.

7. Phenotypic appearance of patients with chromosomal disorders.

8. Methods used in the diagnosis of chromosomal diseases.

9. Etiopathogenesis and clinic of cystic fibrosis.

10. Curation of cystic fibrosis patients.

11. Etiopathogenesis and clinic of hemoglobinopathy (uroximon cell anemia).

12. Curation of patients with hemoglobinopathy (uroximon cell anemia).

13. Etiopathogenesis and clinic of thalassemia.

14. Curation of patients with thalassemia.

15. Enzymopathies: diseases associated with disorders of protein, carbohydrate, nucleotide metabolism.

16. The etiology of galactosemia, pathogenesis.

17. Principles of diagnosis and treatment of galactosemia

18. Lesh-Nayyan syndrome.

19. Prelegial clinic, principles of diagnosis and treatment.

20. Principle of FKU clinic, diagnosis and treatment.

21. The principle of clinic, diagnosis and treatment of alkaptonuria.

22. Clinic, diagnosis and treatment of sphingolipidosis.

23. Clinic, diagnosis and treatment of glycogenosis.

24. Inherited phosphate diabetes. Etiology, pathogenesis.

25. Clinical, diagnostic burns and principles of prevention of hereditary phosphate diabetes.

26. Tony-Debri-Fankoni syndrome.

27. Elers-Danlo syndrome.

28. Principles of medical genetics counseling.

29. Instructions for conducting medical genetics consultation.

30. Family planning.

31. Prenatal diagnosis of hereditary diseases burns.

32. Use of screening methods in diagnostic burns.

33. Describe the methods of medical genetics

34. Etiology, clinic and treatment of cystic fibrosis.

35. Thalassemia etiology, clinic, treatment

36. Clinic, treatment of albinism etiology

37. Shernevsky Turner's disease

38. Conducting prenatal diagnostics

39. Patau disease

40. Consultation in Medical Genetics

41. Etiology, clinic and treatment of Down's syndrome

42. Tubolapathy

43. Monogenic diseases

44. Diseases associated with changes in chromosome number

45. Diseases associated with changes in the structure of chromosomes

46. Shershevsky-Turner syndrome

47. Disorders of gene balance

48. Cat scratch syndrome

49. Dermatoglyphic examination method

50. Immunogenetics test method

51. The method of twins, the purpose of the study.

52. Method of genealogical examination.

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