Lecture complex

Module: "Genes and heredity"

Discipline: Medical Genetics

Module code: GN 1204

Name of EP: 6B10115 "Medicine"

Study hours/ credit hours: 120 hours/4 credits

Course and semester of study: 1-2

Volume of lectures: 3 h.

The lecture complex was developed in accordance with the working curriculum of the EP "Genes and heredity ", the discipline "Medical Genetics" and discussed at the meeting of the department.

Protocol no. __6_ of «_28__» ___12_____ 2023 y.

Head of Department, Professor _____ Yessirkepov M.M.

№ 1

1. Topic: Fundamentals of medical genetics. Research methods of human genetics. Hereditary diseases. Monogenic and polygenic diseases

Chromosomal diseases and their place in general human pathology. Diseases with non-Mendelian type of inheritance.

2. Purpose: To give an idea of the subject and tasks of medical genetics, its role in medicine;

3. Theses of the lecture: Genetics - the science of the laws of heredity and variability. Depending on the object of study, genetics of plants, animals, etc. is classified; depending on the methods used by other disciplines - molecular genetics, ecological and others. Ideas and methods of genetics play an important role in medicine, agriculture, microbiological industry, as well as in genetic engineering.

Medical genetics is the field of genetics, the science that studies:

- phenomena of heredity and variability at all levels of its organisation and existence: molecular, cellular, organismal, population features of manifestation and development of normal and pathological traits,
- role of heredity in human pathology, regularities of transmission of hereditary diseases from generation to generation,
- hereditary human diseases,
- dependence of diseases on genetic predisposition and environmental conditions,
- methods of diagnostics, treatment and prevention of hereditary pathology, including diseases with hereditary predisposition.

Objectives of medical genetics:

- diagnosing hereditary diseases
- analysis of their prevalence in different populations and ethnic groups
- prevention of hereditary diseases on the basis of prenatal (prenatal) diagnostics
- study of molecular-genetic bases of etiology and pathogenesis of hereditary diseases
- identification of sick children
- development of recommendations for their treatment.

It is impossible to study human inheritance using hybridological analysis (crossbreeding method).
For genetic analysis in humans, specific methods are used
specific methods:

- genealogical (method of analysing pedigrees),
- twin,
- cytogenetic,
- biochemical,
- dermatoglyphics and palmoscopy
- molecular-genetic (DNA diagnostics)
- population-statistical,
- somatic cell genetics

Cytogenetic method is based on microscopic study of chromosomes, human karyotype in norm and pathology.

This method makes it possible to establish the presence of human hereditary diseases, to study chromosome structures, to detect translocations, to construct genetic maps, to analyze chromosomal and genomic mutations, to carry out cytochemical studies of gene activity, etc.

The clinical genealogical method was proposed in the late 19th century by F. Galton. It is based on the construction of pedigrees and tracing in a series of generations the transmission of an inherited trait.

The biochemical method makes it possible to determine the contribution of genetic (hereditary) and environmental factors (climate, nutrition, education, upbringing, etc.) in the development of specific traits or diseases in humans.

Hereditary diseases are numerous (over 6000 are known) and diverse in manifestations. Such diseases may be quite rare, but due to the fact that there are many of them, their cumulative frequency is quite high.

They differ from other diseases in that, as a rule, it is possible to find the exact cause of the disease, which is associated with damage to the hereditary apparatus.

Classification of hereditary diseases of man, most commonly used:

1) monogenic mendelian diseases);
2) chromosomal syndromes, resulting from structural or quantitative rearrangements of chromosomes;
3) multifactorial diseases,
4) monogenic diseases with non-traditional, different from Mendelian, type of inheritance - this group was singled out in the last decade.

4. Illustrative material: Overview

https://www.youtube.com/watch?v=0pOYQxa3UCs&feature=youtu.be Mendelian laws
https://www.youtube.com/watch?v=Xh_RplAaNBQ&feature=youtu.be mono, poly, chromosomal diseases

5. Literature: see Annex 1

6. Control questions: (feedback)

1. Who is the father of genetics?
2. What is the importance of genetics to medicine?
3. What characterises independent inheritance?
4. What is the essence of linked inheritance?
5. What are the causes of human hereditary diseases?
6. Can human hereditary diseases be cured?
7. Definition of hereditary diseases
8. Mechanisms of hereditary diseases
9. Monogenic diseases
10. Polygenic diseases
11. chromosomal diseases caused by structural rearrangements of chromosomes: deletions, duplications, inversions, translocations.
12. Definition of the concept of diseases with non-Mendelian type of inheritance.

№ 2

1. Topic: Hereditary diseases.

2. Purpose: Study of hereditary human diseases. Study of etiology, pathogenesis and epidemiology of hereditary diseases

3. Thesis of the lecture: Hereditary diseases are diseases that are transmitted from parents to offspring. Hereditary diseases are formed due to changes in genetic material caused by gene, chromosomal and genogenomic mutations.

Hereditary diseases according to genetic classification are:

* monogenic;
* chromosomal;
* multifactorial (polygenic).

Monogenic diseases are caused by mutations in structural genes in which genetic information is written. Transmission of these diseases to offspring is called Mendelian inherited disease, as it occurs according to the laws of inheritance of H. Mendel. Monogenic type autosomal.- dominant (arachnodactyly, brachydactyly, polydactyly, etc.), autosomal. recessive (more often occurs in persons married to two or sometimes three cousins; agammaglobulinemia, alkaptonuria, etc.) and combining with sex X - and U chromosomes (depending on the gene, the male gets the disease and the disease is carried by the female; haemophilia, etc.). Diseases) are subdivided into hereditary diseases.

Chromosomal diseases are formed due to genomic (change in the number of chromosomes) and chromosomal (change in the structure of chromosomes) mutations. Among the most common chromosomal diseases are trisomies. This is when an additional 3-X chromosome is formed in one of the pairs of chromosomes. For example, autosomes in Down's disease. Trisomy on pairs 21 is present in pairs 13 in Patau syndrome and in pairs 18 in Edwards syndrome. Because of the violation of meiotic division in gametogenesis in women, if there is no one of the sex X-chromosomes, then the syndrome Shereshevsky - Turner, on the contrary, with an excess of one chromosome - leads to the formation of triplo-X syndrome (Klinefelter in men). Chromosomes of infants when carrying a child in women over 35 years of age. Childbirth with this disease is high risk.

Multifactorial diseases result from mutations and interactions of several genes, where adaptations to the disease are increased, and from exposure to environmental factors.
Such diseases include.

- Gout;
- diabetes mellitus;
- hypertension;
- gastric and intestinal ulcers;
- atherosclerosis;
- relates to coronary heart disease, etc.

The cause of this type of hereditary disease has not yet been fully elucidated. Clinical classification of hereditary diseases is carried out according to the organs and systems that have undergone pathological changes. For example, hereditary diseases of the nervous and endocrine systems, circulatory system, liver, kidneys, skin, etc. are classified as hereditary diseases of organs. Clinics and hospitals of neurology, therapy, surgery are engaged in diagnostics and treatment of hereditary diseases in the republic.

4. Illustrative material: Overview

https://www.youtube.com/watch?v=Xh_RplAaNBQ&feature=youtu.be mono, poly, chromosomal diseases

5. Literature: see Annex 1

6. Control questions: (feedback)

1. Who is the father of genetics?
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12. Definition of the concept of diseases with non-Mendelian type of inheritance.

№3

1. Topic: Chromosomal diseases. congenital malformations.

2. Purpose: the role of variability in human pathology; study of genetic mechanisms of chromosomal and specific hereditary diseases, disclosure of congenital malformations.

3. Thesis of the lecture: Chromosomal diseases are formed due to genomic (change in the number of chromosomes) and chromosomal (change in the structure of chromosomes) mutations. Among the most common chromosomal diseases are trisomies. This is when an additional 3-X chromosome is formed in one of the pairs of chromosomes. For example, autosomes in Down's disease. Trisomy on pairs 21 is present in pairs 13 in Patau syndrome and in pairs 18 in Edwards syndrome. Because of the violation of meiotic division in gametogenesis in women, if there is no one of the sex X-chromosomes, then the syndrome Shereshevsky - Turner, on the contrary, with an excess of one chromosome - leads to the formation of triplo-X syndrome (Klinefelter in men). The chromosomes of babies when carrying a child in women over 35 years of age. births with this condition are high risk.

Mechanisms of chromosomal diseases.

The main cause of many chromosomal diseases is a change in the historically, evolutionarily established system - karyotype, that is, a violation of the number of chromosomes or the structure of chromosomes. This is a developmental anomaly, congenital due to a violation of the number of chromosomes and the structure of chromosomes
pathological condition.
It is divided into:
- autosomal syndromes in somatic cells.
- gonosomal syndromes in germ cells.

There are 3 known principles of classification of chromosomal pathology:

1.Characterisation of a known chromosomal mutation.
2.Identification of the type of cells in which the Mutation occurred (gametes, zygote).
3.Identification of past generation mutations.

Clinical picture of chromosomal syndromes:

1. complications in pregnancy-multipregnancy (polyuria)
2. congenital defects in brain development and facial expressions. The size of the head is smaller (microcephaly). The forehead is low .
3. eye cavity-compressed, anophthalmia.
4. tympanic membrane is deformed.
5.cleft upper lip and palate.
6.malformations of internal organs - heart malformations, anomalies of genital organs, kidneys (Keys). uterine appendage, extra spleen.
7.multitasking of fingers and toes.
8.Many children die before one year of age, in the first few days of life (95%).
9.2% of children survive to 10 years of age.
Diagnosis-Chromosomal testing.

4. Illustrative material: Overview

https://www.youtube.com/watch?v=Xh_RplAaNBQ&feature=youtu.be mono, poly, chromosomal diseases

5. Literature: see Annex 1

6. Control questions: (feedback)

1. Who is the father of genetics?
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11. chromosomal diseases caused by structural rearrangements of chromosomes: deletions, duplications, inversions, translocations.
12. Definition of the concept of diseases with non-Mendelian type of inheritance.

5.Literature:

Appendix 1

In Russian:

Basic:

1. Genetics. Textbook for Higher Education Institutions / Edited by Academician of RAMS V.I. Ivanov - Moscow: ICC "Akademkniga", 2006-638c: ill.
2. Muminov T. Fundamentals of molecular biology: a course of lectures. -Almaty: Effekt, 2007.

Additional:

1. Ivanyushkin A.Y., Ignatiev V.N., Korotkikh R.V., SiluyanovaI.V.Izd-vol.
Progress, M.. 2008г.
2. У. Clague, M. Cummings. Fundamentals of Genetics - M.: Technosphere, 2009.
3. Fundamentals of molecular biology of the cell. Textbook. 3 volumes. B. Alberts et al, OZON.RU Publishing House, 2018.

№	Name	Link
1	Electronic library	http://lib.ukma.kz
2	Republican interuniversity electronic library	http://rmebrk.kz/
3	Electronic library of the Medical University "Student Advisor"	http://www.studmedlib.ru
4	"Paragraph" information system "Medicine" section	https://online.zakon.kz/Medicine
5	Scientific electronic library	https://elibrary.ru/
6	Electronic library "BuxMed"	http://www.booksmed.com
7	«Web of science» (Thomson Reuters)	http://apps.webofknowledge.com
8	«Science Direct» (Elsevier)	https://www.sciencedirect.com
9	«Scopus» (Elsevier)	www.scopus.com
10	PubMed	https://www.ncbi.nlm.nih.gov/pubmed

Internet resource:

1. Genetics. Textbook for Higher Education Institutions / Edited by Academician of Russian Academy of Medical Sciences V.I. Ivanov - Moscow: ICC "Akademkniga", 2011-638c: ill.

2. Mushkambarov N.N., Kuznetsov S.N. Molecular biology. Textbook for students of medical universities, 3rd edition, Moscow: Nauka, 2016, 660c.
3. У. Clague, M. Cummings. Fundamentals of genetics - M.: Technosphere, 2009.
4. Kurchanov.A. Human genetics with the basics of general genetics: textbook -SPb, 2009.
5. Alberts B. B., Bray D., Hopkin K. Fundamentals of molecular biology of the cell. Textbook. 2nd ed., revised, per. from Engl. 768st. 2018г.
6. Spirin A.S. Protein biosynthesis, the RHK World and the origin of life.
7. Spirin A.S. Molecular Biology. Structure of ribosomes and protein biosynthesis. - M.: (electronic textbook).

6. Control questions: (feedback)

1. Hereditary diseases. Mechanisms of occurrence.
2. Classification of chromosomal diseases.
3. Chromosomal diseases:
* Monsomia X chromosome syndrome,
* Polysomy X-chromosome syndrome in males and females,
* Y-chromosome polysome syndrome,
* Autosomal monosomy syndrome *
* Autosomal polysomy syndrome,
4. Types of specific (non-traditional) hereditary diseases:
* hereditary diseases in combination with sex
* mitochondrial diseases,
* genomic imprinting diseases,
* trinucleotide repeat expansive diseases,
* prion diseases.
* hereditary metabolic diseases