LESSON № 16

Topic: ROLE OF HEREDITY IN PATHOLOGY

Aim of lesson: to study causes and mechanisms of congenital diseases, there categorization and to meet with phenotypic realizing of congenital pathology.

QUESTIONS:

1. Base of congenital function. Cariotype, genotype, phenotype. Penetrance and expressivity. Factors determining penetrance and expressivity.
2. Congenital diseases.
3. Etiology and pathogenesis of congenital diseases.
4. Mutations and mutates (alcohol, nicotine, radiation e.g.).
5. Categorization of congenital diseases.
6. Genome diseases. The concept of chromosome disorders, their mechanism. Mosaicism. Types of chromosome aberrations: deletion, balanced and Robertsonian translocation, ring chromosome, inversion.
7. Types of genome diseases trait. Sex-chromosomes and autosomes related diseases.
8. Examples of the most common chromosome disorders. Monosomies and trisomies.
9. Biochemical and molecular basis of single-gene (Mendelian) disorders.
10. Features of disorders and examples of disorders transmitted by
    - autosome-dominant trait;
    - autosome-recessive trait;
    - sex chromosome-linked (X-linked) trait
11. Gene diseases:
    - metabolic diseases (glycogenosis, phenylketonuria, galactosemia e.g.);
    - blood diseases (hemoglobinosis S, elliptocytosis, hemophilia e.g.).

12. Investigative methods for congenital diseases:
    - genealogical;

- population;
- statistical;
- cytogenetical;
- biochemical;
- dermatoglyphical.

13. Features of multifactorial (polygenic) disorders. The most common multifactorial disorders.
14. Diseases of congenital supports.
15. Embryopathic and fetopathic disorders. The critical periods of pregnancy. Phenocopies.
16. Medical genetic consulting.
17. Constitution. Types of constitution.

Tasks

1

N., a healthy woman, visited a genetic counseling unit for consultation. She told her doctor that her father suffered from color blindness, but her mother had no vision problems. N. was anxious about the risk of color blindness to her future children.

What are the manifestations of color blindness and the pattern of its inheritance?

Can N. carry the gene of color blindness in her genotype?

What is the probability of disease and the probability that her children (separately boys or girls) will be the carriers of the abnormal gene?

2

N., a healthy woman, whose father suffered from hemophilia A and mother was healthy, went to a genetic counseling unit. She was anxious about the risk of hemophilia to her grandsons. Her husband, as well as her son and two daughters were healthy.

What is the pattern of inheritance of hemophilia A? What are the main features of this type of inheritance?

What is the risk of the disease descending from her son to her grandson and from her daughter to the grandchildren?

What are the etiology and pathogenesis of hemophilia A?

Can this disease have lethal and sublethal forms?

3

Patient S., a pregnant woman, went to a genetic counseling unit for consultation. She told her doctor that her sister suffered from phenylketonuria. In her husband's pedigree there are marriages between close relatives, but none of the children have had phenylketonuria. A thorough examination of the patient and her husband revealed no signs of pathology.

How great is the risk of phenylketonuria to patient S.'s sons?

What are the etiology and basic mechanisms of phenylketonuria? Is gender significant for its inheritance?

What are the main manifestations of the disease and their pathogenesis?

What is the approach to early diagnostics of phenylketinuria in the newborns?

Is it possible to prevent phenylpyruvate oligophrenia in children?

4

A male patient, 3 years old, was admitted to the pediatric department. On examination: signs of growth retardation, flat facial profile, half-open mouth, oblique palpebral fissures and epicanthic folds are evident; transverse skin folds on the palms. A study of the patient's karyotype revealed the following: 46, XY, + t (+14, 21).

What disease does the patient suffer from?

Describe and characterize the patient's karyotype? By what features does it differ from the normal one?

What are the possible causes and pathogenesis of this disease?

Are other variants of karyotype modification possible in this disease? Which of the variants is the most common?

5

Patient M., male, 21 years old, suffers from hereditary form of hypogammaglobulinemia. His father also suffers from this disease; mother is healthy. An examination showed a significant decrease of IgM and IgG levels in the patient's blood.

What is the pattern of its inheritance?

Characterize the genotype of the patient's mother with respect to this pathology, and also the possible genotypes of the patient's siblings. How much is the risk for the patient's sibs to develop hypogammaglobuliemia?

LITERATURE:

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