DIAGNOSIS AND TREATMENT OF HEREDITARY DISEASES OF THE CELL FUND

Madaminov Muxammadvali Ikromjon o‘g‘li
Tashkent Medical Academy

https://doi.org/10.5281/zenodo.7219598

Abstract. *Hereditary diseases are diseases caused by a violation of genetic information (genetic information); It is usually caused by mutations in chromosomes or genes and is passed down from generation to generation. Mutations can be caused by environmental factors (ionizing radiation, some biologically active chemical compounds) and adverse effects on organisms and cells. This article will give you more information!*

Kezwords*: Heredity, disease, gene, cell, dominant, recessive.*

ДИАГНОСТИКА И ЛЕЧЕНИЕ НАСЛЕДСТВЕННЫХ ЗАБОЛЕВАНИЙ КЛЕТОЧНОГО ФОНДА

Аннотация*. Наследственные болезни – это заболевания, вызванные нарушением генетической информации (генетической информации); Обычно это вызвано мутациями в хромосомах или генах и передается из поколения в поколение. Мутации могут быть вызваны факторами окружающей среды (ионизирующее излучение, некоторые биологически активные химические соединения) и неблагоприятными воздействиями на организмы и клетки. Эта статья даст вам больше информации!*

Ключевые слова: *Наследственность, болезнь, ген, клетка, доминантный, рецессивный.*

INTRODUCTION

Inheritance and hereditary diseases

Hereditary diseases - diseases caused by a violation of genetic information (genetic information); It is usually caused by mutations in chromosomes or genes and is passed down from generation to generation. Mutations can be caused by environmental factors (ionizing radiation, some biologically active chemical compounds) and adverse effects on organisms and cells.

MATERIALS AND METHODS

I. to. It is studied mainly by clinical and genealogical methods, in which genealogy is formed. Using this method, I. to. (autosomal dominant, autosomal recessive and sex-linked diseases) can be inherited in various ways. In autosomal dominant diseases, the disease is controlled by dominant genes located on autosomes. The disease occurs in more than 50% of cases in each generation. Brachydactyly, arachnodactyly, retinoblastoma, some types of psoriasis, etc. in this way it is transmitted from generation to generation.

Autosomal recessive I.k. If parents have pathological recessive genes, there is a risk of having a sick child, so these diseases do not occur in every generation. However, this probability increases when a family is formed between close relatives with altered genes. These include phenylketonuria, myoclonia, epilepsy, some types of oligophrenia, etc. may be an example.

Specific autosomal dominant and autosomal recessive I. to. (for example, some types of color blindness, hemophilia A, sideroahrestic anemia, etc.) are inherited by gender.

RESULTS

The twin method is also used to study certain features of hereditary traits. Twins are known to have one or two eggs. Identical twins are characterized by the same genotype and

appearance (phenotype), as well as homogeneity. The genotypes of identical twins are different and differ from each other in appearance and sex. The twin method does not study the laws of heredity of certain diseases, but the study of certain I. to. also allows you to determine the tendency to

The chromosome and the I. k.i gene are different. Chromosomal disorders are mainly due to changes in the structure and number of chromosomes, which account for 1% of newborns. Significant changes in chromosomes often limit the vital activity of the organism and lead to the death of the developing fetus. These diseases are caused by changes in autosomes and sex chromosomes. These include such syndromes and diseases as Shereshevsky-Turner (caryoty-pi-XO), Klinefelter (XXY), Patau (trisomy 13), Down, "cat's cry".

In general, many chromosomal disorders include changes in the structure of the human skeleton and nervous system, congenital malformations of the external and internal organs, short stature, nervous, endocrine, etc. systemic disorders are observed, the generative activity of patients decreases.

Gene diseases are associated with metabolism resulting from point mutations. To date, more than 30 species have been identified. Thus, disorders of fat metabolism are accompanied by changes in the activity of the central nervous system. The most severe of these is the Tay-Sachs amavrotic idiocy, in which there is a decrease in visual acuity, dementia, etc. neurological symptoms are observed.

Hereditary diseases: what are they?

Heredity is a set of hereditary diseases, i.e. diseases and ailments that tend to be transmitted from parents to children through the transfer of genes. calls them.

Thus, these are diseases that occur at the chromosomal, mitochondrial or Mendelian level, where there are genetic mutations inherited from our ancestors. It is not always necessary for one of the parents to exhibit the disorder or disease, depending on the type of inheritance that is manifested: it may be the carrier of a recessive gene that does not trigger the onset of the disease. he or she, but may develop in these generations.

It is important to note that genetic diseases and hereditary diseases are certainly not synonymous. And yet, although all hereditary diseases are genetic, the truth is that the reverse relationship does not always have to occur: de-novo, which occurs without a family history, genetic diseases arise as a result of spontaneous mutations.

For a disease to be hereditary, the genes and mutations associated with its occurrence must be present in the germ cells, that is, in the sperm and/or eggs that form part of the new organism. Otherwise, we are faced with a hereditary disease, not a hereditary one.

DISCUSSION

Types of gene distribution

It is necessary to consider the many ways of genetic transmission that a mutated gene can transmit in order to know where speech and hereditary diseases come from. In this sense, some of the main methods of genetic transmission are as follows.

Types of gene distribution

It is necessary to consider the many ways of genetic transmission that a mutated gene can transmit in order to know where speech and hereditary diseases come from. In this sense, some of the main methods of genetic transmission are as follows.

CONCLUSIONS

1. **Autosomal dominant inheritance**

One of the main and best known types of inheritance is autosomal dominant inheritance, in which a mutation occurs on one of the non-sex or autosomal chromosomes. The dominant gene is always the gene that is expressed, so if it has a mutation associated with the onset of the disease, it will show up and develop.

At the same time, each child has a 50% chance of developing the disease (depending on who inherited the dominant gene). It can have complete penetration (one allele predominates over the other) or incomplete (two dominant genes are inherited, inherited traits are a mixture of what is inherited from the parent).

2. Autosomal recessive type of inheritance.

Autosomal recessive inheritance is when a recessive gene mutates or changes and it is passed on to the next generation. Now the fact that this change is in a recessive gene means that the disease will not develop until it is present in several alleles of the chromosome. the presence of a copy of this gene does not mean that a disease must occur.

REFERENCES

1. Children's_diseases_8_th_edition_2_volume.pdf
2. Children's_diseases_8_th_edition_1_volume.pdf
3. Neonatology. Volume 2.pdf
4. UMUMIY_AMALIYOT_SHIFOKORI_CHUN_PEDIATSIA.doc
5. Nelson Pediatrics. Volume 4 2009.pdf
6. Neonatology_National_guideline.pdf
7. Basic_help_to_newborn,_Volodin.PDF
8. Nelson Pediatrics. 2 vol. 2009.pdf
9. Qizi T. J. I., Farrukh S. TREATMENT OF MYOCARDIAL INFARCTION AND FIRST AID //Science and innovation. – 2022. – Т. 1. – №. D3. – С. 317-320.
10. Shernazarov F., Azimov A. INCREASED BRAIN PRESSURE-CAUSES, SYMPTOMS, COMPLICATIONS, TREATMENT //Современная медицина: новые подходы и актуальные исследования. – 2021. – С. 73-77.
11. qizi Tohirova J. I., og‘li Ibragimov B. I., og‘li Shernazarov F. F. CONGENITAL HEART DISEASE-CAUSES, CLASSIFICATION, DIAGNOSIS, TREATMENT, COMPLICATIONS, CONSEQUENCES //Eurasian Journal of Medical and Natural Sciences. – 2022. – Т. 2. – №. 3. – С. 84-89.
12. Mratbaevna W. N., Farrux S. The Structure of the Heart and its Physiology in Regular Athletes //Eurasian Scientific Herald. – 2022. – Т. 8. – С. 102-105.
13. Farhod o’g’li S. F. GASTRIT—SABABLARI, ALOMATLARI, TASHXISLASH, DAVOLASH, DORILAR, ASORATLARI, OLDINI OLISH //Лучший инноватор в области науки. – 2022. – Т. 1. – №. 1. – С. 103-107.
14. Фаррух Ш. и др. ПУТИ УСТРАНЕНИЯ САХАРНОГО ДИАБЕТА //Science and innovation. – 2022. – Т. 1. – №. D3. – С. 313-316.