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ЗАБОЛЕВАНИЯ С X-ДОМИНАНТНЫМ ТИПОМ НАСЛЕДОВАНИЯ

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DISEASES WITH AN X-DOMINANT TYPE OF INHERITANCE

Аннотация. Данная статья посвящена заболеваниям с х-доминантным типом наследования, особенностям этих заболеваний и их лечению. Главная характеристика X-сцепленного доминантного наследования заключается в том, что больные мужчины передают аномальный ген (или заболевание) всем своим дочерям и не передают его сыновьям. Больная женщина передает X-сцепленный доминантный ген половине своих детей независимо от пола. Цель данной статьи – рассмотреть особенности заболеваний, сцепленные с X-хромосомой и выявить отличительные черты этих заболеваний у женского и мужского пола.

Ключевые слова. Наследование, ген, заболевание, доминантный

Abstract. This article is devoted to diseases with x-dominant type of inheritance, peculiarities of these diseases and their treatment. The main characteristic of X-linked dominant inheritance is that sick men transmit an abnormal gene (or disease) to all their daughters and do not pass it on to their sons. A sick woman transmits the X-linked dominant gene to half of her children regardless of gender. The aim of this article is to examine the characteristics of X-chromosome-bound diseases and to identify the distinctive features of these diseases in both males and females.

Key words. Inheritance, gene, disease, dominant

Introduction

Currently, more than 370 diseases associated with X-chromosome have been described. The degree of manifestation of symptoms depends on the sex of the body. As a rule, the full form of the disease occurs mostly in men, because they are homozygous organisms on the genes located on the X-chromosome.

A mutational change may affect the recessive gene linked to the X chromosome. In this case, the disease is called XR disease. In this case, heterozygous women do not show signs of pathology, but they may inherit it. Homozygotes in most cases are lethal.

In the case of mutations of the dominant gene located on the X chromosome, the XD disease is spoken of. In this case, it appears in heterozygous women in mild form. Homozygous organisms are also usually lethal.

X-linked diseases cannot be transmitted from father to son in principle. At birth, the boy receives a Y chromosome from his father, not his X chromosome with an abnormal version of the gene. On the other hand, when a daughter is born, the disease is always transmitted, making the child at least a carrier of mutation.

Dependence on the location of the gene

In the case of autosomal diseases, the pathology can be transmitted to all children without exception. In cases of gender-bound inheritance, this is not the case because the gene is not on the autosome.

When a gene is found in a Y-chromosome, its encoded feature (or pathology) can only appear in the heterogamous sex (i.e., in all male children). Due to the very small number of genes present on the Y chromosome, the vast majority of sex-disconnected genes are associated with the X chromosome. Some sexual differentiation disorders are linked to the Y chromosome, because it is on this chromosome that a special SRY gene is located, which determines the testicular development and the organism's belonging to the male sex.

At the same time, when the gene comes into contact with an X chromosome, inheritance is more complicated. Thus, dominant versions of mutations are more often manifested in the homogeneous field, which in humans is a female sex. Recessive changes, on the contrary, will be more noticeable in organisms with heterogeneous (male) sex. The reason for such irregularity is that the recessive gene requires two copies of the modified gene for manifestation in the XX genotype, whereas in the XY-genotype, the recessive gene does not have an overwhelming normal version, so the corresponding symptoms appear more easily.

The mechanism of inactivation of one of the X chromosomes should also be taken into account. With normal body development, only one X chromosome remains from each pair of X chromosomes. Accordingly, if the version with the anomalous gene is inactivated, even the dominant feature will not appear.

Examples of X-linked inheritance include color blindness and hemophilia. Both of these diseases are recessive, with the corresponding genes in the X chromosome. Statistics confirm that such diseases are more common in men than in women. In addition, if there is a single copy of the recessive gene, the woman remains healthy, but is a carrier of the disease.

Manifestations of XD disease

These diseases are less common than XR pathologies. At the same time, they are very likely to cause intrauterine male fetal death with a mutant homozygosity. Therefore, some diseases that are also accompanied by frequent spontaneous interruptions of pregnancy are considered to be caused by XD defects (e.g. focal mesoectodermal dysplasia and Franceschetti-Yadasson syndrome).

If a woman has a homozygous dominant non-lethal gene, all her children will receive one version of the mutational change. For a heterozygous case, the mutant gene will be inherited by half of the children, regardless of gender. Homozygous men can pass on such allele only to their daughters (without exception). This fact explains the observed statistics: if there is a non-lethal XD defect in the family, the prevalence of sick women will be observed.

Endocrine XD diseases include certain forms of pseudohypoparathyreosis and nephrogenic diabetes mellitus.

Treatment and prenatal diagnostic capabilities

Some diseases whose inheritance is linked to the X chromosome can be successfully diagnosed, and you can even find out the heterozygosity of the parent. Treatment methods can also be created for them. If it is not possible to accurately determine the genetic defect (e.g. because there is no appropriate molecular probe available), testing is usually limited to determining the sex of the child. If the embryo is a boy, the probability of developing the disease for him is 50 percent (provided that the parents are at risk for XR disease). In such a case, it is up to the parents to decide whether to terminate or retain the pregnancy.

Difficulty in distinguishing between dominant and recessive forms

X-linked chromosome dominant and recessive inheritance refers only to the way in which pathological change manifests itself in women. However, due to the accidental inactivation of one of the chromosomes in the cells of the female body, the boundary between these two variants is blurred. For this reason, heterozygous organisms may have signs of abnormalities. Accordingly, this group of diseases is often described simply as linked to an X chromosome, without indicating the dominant or recessive nature of inheritance.

However, heterozygous organisms are healthy in some female-bound diseases (e.g. Gunther's syndrome). In this case, separation makes sense. Similarly, it is appropriate to talk about the type of inheritance for diseases in which female heterozygous organisms show the same signs of pathology as homozygous men, as is the case with X-linked hypophosphatemia rickets.

Disease peculiarities

Affected men married to a healthy woman have no sick sons or healthy daughters.

Both sons and daughters of women carriers have a 50% risk of inheriting a phenotype. Genealogy is similar to autosomal dominant inheritance.

Affected women occur almost twice as often as men, but usually have variable but milder manifestations of the phenotype.

Conclusion

The general conclusion is that illnesses inherited recessively linked to the X chromosome are almost exclusively in boys; they are not transmitted from father to son, but are inherited from mother. Diseases that are dominant and X-linked can be observed in both boys and girls, and are not transmitted from father to son.

Diseases characterized by X-linked dominant inheritance include vitamin-D-resistant rickets (rickets that cannot be treated with normal doses of vitamin D), oral-facial-finger syndrome (multiple hyperpolarized frenulum's of the tongue, clefts of the lip and palate, hypoplasia of the wings of the nose, asymmetric shortening of the fingers) and other diseases.

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