HEREDITARY DISEASES AND THEIR TYPES

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Abstract This article provides information about the types of genetic diseases, their origin, ways to deal with them and their detrimental consequences. In this article, we can also see our specialists who are currently fighting such diseases.

Key words: Hereditary diseases, Autosomal recessive inheritance, Polygenic inheritance, Mitochondrial inheritance,Cystic fibrosis, Phenylketonuria, Leber's congenital amaurosis.

Introduction

Over time, such a science as medicine has become more and more developed, this thing is life expectancy made it possible to significantly increase the duration, iys quality and well-being. Due to the fact that today most of the diseases that once led to a dead state it can be successfully treated, in some cases the disease itself has also been eliminated.That's despite the fact that it causes a big problem for medicine, such as AIDS, cancer or diabetes there are different types of diseases. In addition to them, by our ancestors a large group of diseases that are associated with genes that are infected and mainly do not have a cure avaible (although sometimes reducing or slowing down or correcting the symptom, even if reducing treatments are found or occur in the subject and in the daily life of your partner destroy the establishment that comes). We are talking about a set of hereditary diseases, we are talking about this article a concept that reflects during. Hereditary diseases: what are they?

A set of hereditary diseases is called lineage, that is from parents to children, infection of genlam diseases and diseases that have the property of being transmitted

through, causes them. Thus, it is the one that appears at the level of the chromosome, mitochondrial and the father-from our grandfathers it comes from our ancestors diseases in which there are genetic mutations. From parents showing a disorder or illness depending on the type of inheritance that is a payment for one each always number is not a requirement: maybe it will not trigger the onset of disease regressive gene can be a carrier, he or she, but can develop in these generations. It is important to pay attention to genetic diseases and hereditary diseases, of cource, synonymous not. And even though hereditary diseases are genetic, the truth is that the reverse relationships do not have to arise all the time: where our family history has appeared, our own-mutations that arise from are caused by genetic diseases present.

In order for the disease to be hereditary, genes that are associated with it's appearance and mutations in thegincial cells, that is, the sperm that made up part of the New being, and or should be in ovules. Otherwise we will face a hereditary disease, not hereditary.

In the types of distribution of genes, to find out where hereditary diseases come from many methods of genetic transmission in which a mutated gene can be sent it is necessary to take into account. In this context, the main methods of genetic transmission are as follows.

1.Autosomal dominant inheritance. The main and best known of the inheritance one of the types is autosomal dominant inheritance, in which it is asexual or autosomal there is a mutation in one of the chromosomes. The Dominant gene will always be the gene to be expressed, therefore, if there is a mutation in it, which is associated with the appearance of the disease, then it itself manifests and develops. In this case, the manifestation of the disease in every child the probability will be 50% (depending on who inherited the dominant gene). It has a full penetration of can have (one allele dominates the other) or incomplete (two domimamt genes inherited, inherited properties are something that comes from your parents mix).

2.Autosomal recessive inheritance. Autosomal recessive inheritance is a mutation in a recessive gene or it appears when there is an alternation, and this is

passed on to a new generation. Now this change is recessive the fact that it is in the gene is a disease until it is present in several alleles of the chromosome means that it does not develop having a copy of this gene disorder should occur does not mean. For both alleles of the gene for this to occur, the mutation is it will have to show, that is, the father and mother modified the gene for the development of the disease they need to deliver a copy to the child.

3.Sex-related heredity. They are added to the sex cells for infection although necessary, most hereditary diseases are autosomal, that is, the change is sexual tosmoke chromosome is present in one . But other diseases of the sex chromosome transmitted through copies, X or Y. Only at the genetic level do men have Y chromosomes carry, if there are changes in this chromosome, it is only from parents to male children can be transmitted. In the event that the change occurs on the X chromosome, regardless of gender they are both parents can infect their children.

4.Polygenic Inheritance. The two previous types of genetic inherintance are monogenic, that is, to one gene related. Nevertheless, there are several that are often associated with the onset of the disease there are genes. In this case, we will talk about polygenic inheritance.

5.Mitochondrial heritage. Although they are as well known or common as the previous ones although not, it is a variety of Hereditary that does not originate from the DNA present in chromosomes there are diseases and disorders, but it’s origin is the so-called mitochondria in organoids. In these structures we can also find DNA, although in this case it is only if it comes from a mother.

So let’s give examples of hereditary diseases. Finding thousand of diseases. Finding thousands of diseases there are many possible hereditary diseases. However, to some hereditary diseases face and name to put it, below we leave you a total of dozens of examples (some of which are well-known).

1.Huntington’s disease. Huntington’s disease, formerly known as Huntington’s chorea, hereditary disease with complete penetrative autosomal dominant transmission. Other symptoms of this disease the line that causes movement changes

(involuntarily your muscles when moving emphasizing the choreological action they perform due to their contraction), as well as cognitive function, and especially by inducing a profound change in function controllers excretion is characterized by progressive neurodegeneration, deteriorates over time.

2.Hemophilia. Characterized by difficulties in blood coagulation and abundant and this dangerous disease that causes continuous bleeding is life-threatening if not stopped can give birth, it is also a hereditary disease. In particular it's most common form, A type hemophilia is a disease associated with sex chromosomes (especially with the X chromosome tied) and relayed recessive. Therefore, hemophilia is almost exclusively male it is a disease that is damaged by, since the appearance of the X chromosome in women there are two copies that will be difficult.

3.Achondroplasia. Achondroplasia is a genetic disease that is the main cause of dwarfism is characterized by causing changes in cartilage and bone formation. Although in most cases (about 80%) we are with mutations that arise from by self even if we deal with it, the presence of a family history in which 20% of them inherited a mutation observed. In such cases , an autosomal dominant pattern is observed, in which the mutated can cause a cup of the gene disease (if one of the parents has this disease if infected, their children have a 50% change of developing acondroplasia). Main related genes are G1138A and G1138C.

4.Marfan's disease. Disease of genetic origin impact on connective tissue characterized by making. It is an autosomal dominant disorder in which the bones are uncontrolled increases in size, in addition to other symptoms such as cardiovascular (aortic noises and can be life-threatening by highlighting the effects) or eye level (Retinal cleavages, may be myopia) and cataracts).

5.Cystic fibrosis. Cystic fibrosis is caused by autosomal recessive inheritance exiting and characterized by the accumulation of mucus in the lungs, breathing it is one of the hereditary diseases that make it difficult. Sputum is also found in organs such as the pancreas can appear, cysts can also appear on them. It is usually severely

contagious it is a life-threatening disease caused by diseases, often in children and young people it is found.

6.Ley syndrome. In this case, we are faced with a hereditary disease of the mitochondrial type we are coming (although it can also occur in autosomal recessive genetic transmission) characterized by rapid neurodegeneration that occurs early (usually from the first year of life before) and in which damage is found to the brainstem and basal ganglia. Hypotonia, movement and walking problems, respiratory problems, neuropathy, heart, kidney and common symptoms such as OPCA's work disorder.

7.Sickle cell anemia. This disorder is characterized by the presence of changes in The Shape of red blood cells (to become round, they are irregular in shape has it hardens), thus ensuring the blockage of blood flow, as well as,to the reduction of the life of the Globes mentioned above (which means a decrease in your levels thing) very necessary for this ingredient blood). It is another through autosomal recessive inheritance hereditary disease.

8.Thalassemia. Associated with inherited blood through recessive autosomal inheritance another disease is Thalassemia. Synthesis of certain parts of hemoglobin in this disease causes difficulty in making (especially alpha globin), smaller amounts of red blood cells causing anemia, which leads to its appearance and even to varying degrees and severity what comes out (although with treatment they can led a normal life)

9.Duxenne muscular dystrophy. Progressive muscle weakness (also voluntary, both involuntary at the muscle level), frequent falls, constant fatigue and sometimes characterized by intellectual disability, this serious degenerative disease is fundamentally hereditary, X is a recessive inheritance pattern associated with the chromosome.

10.Phenylketonuria. Phenylketonuria is a hereditary disease, through autosomal recessive inheritance is obtained and with this characterized by absence or lack of phenylalanine hydroxylase, citing the inability to break down

phenylalanine in such a way that it accumulates in the body what comes out. This can lead to brain damage and it is usually caused by maturation delay, intellectual incompetence, uncontrolled movements and even seizures, as well as causes a specific smell of urine and sweat.

11.Leber is a congenital Amaurosis. Abnormalities of photoreceptors in the retina or progressive a rare disease characterized by degeneration. Great to make your eyesight worse can cause vision disorders and see those who suffer from it is common for the ability to be very limited. It is inherited in an autosomal recessive manner past illness.

In conclusion, the main one that stands before our medicine in the present period one of the tasks is harmful consequences from marriages between close relatives, complications are considered. How many families have children under the influence of such harmful consequences are suffering in the form of disabilities. To prevent diseases of this type, medical and genetic consultations, reproductive centers, sklir centers are functioning.

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