Your Name

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Date

Gene Mutations

A gene mutation is a permanent alteration of the DNA sequence from which a gene is composed. The size of the mutations varies, which affects from a single basic component (base pair) of the DNA to a large segment of a chromosome with several genes. In genetics, a spontaneous and unpredictable variation in the sequence of genes that make up the DNA of a living being is called, which introduces specific changes of a physical, physiological or other type in the individual, which may or may not be inherited from His descendants.

Mutations can translate into positive or negative changes in the vital performance of organisms, and in that sense they can boost adaptation and evolution (even creating new species in the long run), or they can become genetic diseases or inherited defects (Solinas, Cinzia, et al). These types of changes occur for two essential reasons: spontaneously and naturally, due to errors in genome replication during the phases of cell replication; or externally, due to the action of mutagens of various types on the organism, such as ionizing radiation, certain chemical substances and the action of some viral organisms, among others.

Most of the mutations that living beings experience are of a recessive type, that is, they do not become manifest in the individual in which they originate, but remain inactive and inadvertent, although they can be transmitted to the offspring in case they (at least for multicellular living beings) the alteration occurs in the content of sex cells (gametes).

Mutations can occur at three levels:

Molecular (gene or specific) . They occur in the chemical bases of DNA, that is, in their own nitrogen bases, due to some change in the fundamental elements that compose them.

Chromosomal. A segment of chromosome is altered, that is, much more than a gene, and in that sense large amounts of information can be lost, duplicated or relocated (Pearson, 400)

Genomic. It affects a certain set of chromosomes, causing excesses or lack thereof, and substantially varying the entire genome of the organism.

As it turned out, there are special changes that accumulate during the life of generations. It also became known that there are facial mutations, consisting in a slight distortion of the original product. The provision on the re-emergence of new biological traits applies exclusively to gene mutations.

Causes of Gene Mutations

Mutagenic factors. Most mutations have a detrimental effect on the body, disrupting traits regulated by natural selection. Each organism is prone to mutation, but under the influence of mutagenic factors, their number increases dramatically. These factors include: ionizing, ultraviolet radiation, elevated temperature, many compounds of chemicals, as well as viruses.

Antimutagenic factors, that is, protection factors of the hereditary apparatus, can safely be attributed to the degeneracy of the genetic code, the removal of unnecessary sites that do not carry genetic information (introns), as well as the double chain of the molecule’s DNA.

1. Progeria . Progeria is considered one of the rarest genetic defects. This mutation manifests itself in premature aging of the body. Most patients die before they reach the age of thirteen, and few manage to save their lives up to twenty years. This disease develops strokes and heart disease, and that is why, most often, the cause of death is a heart attack or stroke.

2. Syndrome on Juner Tan (SUT). This syndrome is specific in that those exposed to it move on all fours. Usually people with SUT use the simplest, most primitive speech and suffer from congenital cerebral insufficiency.

3. Hypertrichosis. Also has the name “werewolf syndrome” or - “Abrams syndrome”. This phenomenon has been traced and documented since the Middle Ages. People prone to hypertrichosis differ in body hair in excess of the norm, especially for the face, ears and shoulders.

4. Severe combined immunodeficiency. Those affected by this disease already at birth are deprived of an effective immune system, which the average person possesses. David Vetter, thanks to whom the disease became famous in 1976, died at the age of thirteen, after an unsuccessful attempt at surgical intervention to strengthen immunity (Gataullina, Svetlana, et al. 389).

5. Marfan syndrome. The disease occurs quite often, and is accompanied by a disproportionate development of the limbs, excessive mobility of the joints. A deviation expressed by intergrowth of ribs is much less common, resulting in either bulging or sinking of the chest. A common problem prone to bottom syndrome is spinal curvature.

To conclude, it is important to understand that determining how harmful or beneficial it is depends largely on the genotypic environment. Many environmental factors can disrupt the ordering of genes, a strictly established process of their self-reproduction. In the process of evolution and natural selection, man acquired not only useful features, but also not the most favorable ones related to diseases. The human species pays for what it received from nature through the accumulation of pathological signs.

Works Cited

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