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Commentary

Disease Causation and Beneficial Mutations

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DESCRIPTION

Mutations are changes in the nucleotide sequences of the genome of an organism, virus, or extra chromosomal DNA. The viral genome contains either DNA or RNA. Mutations are caused by errors during DNA or virus replication and by other types of DNA damage such as pyrimidine dimers which are caused by mitosis or meiosis, or exposure to UV light and are error-prone repairs. It can be affected or cause failure in other forms of repair. An error occurred during replication. Mutations can also result from the insertion or deletion of DNA segments by mobile genetic elements.

DISEASE CAUSATION

Changes in DNA caused by mutations in the coding regions of DNA can cause errors in protein sequences, resulting in partially or completely dysfunctional proteins. To function properly, every cell relies on thousands of proteins that function in the right place and at the right time. When mutations change proteins that play important roles in the body, they can lead to medical conditions. Studies comparing genes between different species of Drosophila have shown that if mutations alter proteins, mutations are probably harmful, with an estimated 70% of amino acid polymorphisms having adverse effects, the rest being neutral or weak. It suggests that it is beneficial. Some mutations alter the DNA sequence of a gene, but not the protein produced by the gene. Studies have shown that only 7% of point mutations in yeast non-coding DNA are harmful and 12% of coding DNA are harmful. The remaining mutations are neutral or slightly beneficial.

Inherited disorders

If a mutation is present in germ cells, this can lead to a fault that carries a mutation in whole cells. This is in the case of hereditary disease. In particular, if there is a mutation in the DNA repair gene in germ cells, people who carry such germline mutations may increase cancer risk. The list of such germline mutations of 34 is identified in the article DNA repair disorder. One example is a mutation that occurs in Albinism, *OCA1* or *OCA2* gene. Individuals with this disorder are often many types of cancer and other obstacles, and visual impairments have occurred.

DNA damage can cause errors when DNA is replicated, and this replication error can cause genetic mutations, which in turn can lead to genetic damage. DNA damage is repaired by the cell's DNA repair system. Every cell has a set of signaling pathways that enzymes use to detect and repair DNA damage. The process of DNA repair is an important method that the body uses to protect itself from illness, as DNA can be damaged in many ways. After DNA damage causes a mutation, the mutation cannot be repaired.

Role in carcinogenesis

Mutations, on the other hand, can occur in the somatic cells of an organism. Such mutations are present in all progeny of that cell within the same organism. Accumulation of specific mutations over generations of somatic cells is part of the cause of malignant transformation from normal cells to cancer cells.

Cells with heterozygous loss-of-function mutations (one good copy of the gene and one mutated copy) function normally with the unmutated copy until the good copy spontaneously somatically mutates. I can do it. This type of mutation is common in living organisms, but its proportion is difficult to measure. Measuring this rate is important in predicting the rate at which people are likely to develop cancer.

Prion mutations

Prions are proteins and do not contain genetic material. However, prion replication has been shown to be affected by mutation and natural selection, as well as other forms of replication. The human PRNP gene encodes the major prion protein, PrP and is affected by mutations that can lead to disease-causing prions.

BENEFICIAL MUTATIONS

Mutations that cause changes in protein sequences can be harmful to the organism, but their effects can be beneficial in

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certain circumstances. In this case, the mutation may allow the mutated organism to withstand certain environmental stresses or reproduce faster than wild-type organisms. In these cases, mutations become more common in the population by natural selection. Examples include:

HIV resistance

Deletion of a specific 32 base pair of human CCR5 (CCR5Δ32) confers HIV resistance to homozygotes and delays the development of aids in heterozygotes. A possible explanation for the relatively high frequency of CCR5Δ32 in the European population is that it conferred resistance to bubonic plague in Europe in the mid-14th century. People with this mutation were more likely to survive the infection. Therefore, its frequency in the population has increased. This theory can explain why this mutation is not found in southern Africa, which has escaped bubonic plague. More recent theories suggest that the selective pressure for the CCR5 delta 32 mutation was caused by smallpox rather than bubonic plague.

Malaria resistance

An example of a harmful mutation is sickle cell anemia. This is a blood disorder in which the body produces an abnormal type of oxygen-carrying substance hemoglobin in red blood cells. One-third of all indigenous peoples in sub-Saharan Africa carry alleles. In areas where malaria is common, carrying only one sickle cell allele is essential for survival. People who carry only one of the two allogeneic genes for sickle cell disease are more resistant to malaria because the disease of the infected cells stops the transmission of plasmodium.

Antibiotic resistance

When exposed to antibiotics, virtually all bacteria develop antibiotic resistance. In fact, the bacterial population already has such mutations that are selected during the selection of antibiotics. Apparently, such mutations benefit only the bacteria, not the infected ones.

Lactase persistence

The mutation allowed humans to express the enzyme lactase and adults to digest lactose after being naturally separated from breast milk. This may be one of the most beneficial mutations in human evolution in recent years.