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Mutation: A genetic change

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DESCRIPTION

Mutation, an alteration in the genetic material (the genome) of a cell of a living organism or of a virus that is more or less permanent and that can be transmitted to the cell's or the virus's descendants. (The genomes of organisms are all composed of DNA, whereas viral genomes can be of DNA or RNA; see heredity: The physical basis of heredity.) Mutation in the DNA of a body cell of a multicellular organism (somatic mutation) may be transmitted to descendant cells by DNA replication and hence result in a sector or patch of cells having abnormal function, an example being cancer. Mutations in egg or sperm cells (germinal mutations) may result in an individual offspring all of whose cells carry the mutation, which often confers some serious malfunction, as in the case of a human genetic disease such as cystic fibrosis. Mutations result either from accidents during the normal chemical transactions of DNA, often during replication, or from exposure to high-energy electromagnetic radiation (e.g., ultraviolet light or X-rays) or particle radiation or to highly reactive chemicals in the environment. Because mutations are random changes, they are expected to be mostly deleterious, but some may be beneficial in certain environments. In general, mutation is the main source of genetic variation, which is the raw material for evolution by natural selection.

An individual offspring inherits mutations only when mutations are present in parental egg or sperm cells (germinal mutations). All of the offspring's cells will carry the mutated DNA, which often confers some serious malfunction, as in the case of a human genetic disease such as cystic fibrosis. Mutations in DNA occur for different reasons. For

example, environmental factors, such as exposure to ultraviolet radiation or certain chemicals, can induce changes in the DNA sequence. Mutations can also occur because of hereditary factors. Mutation hotspots (or mutational hotspots) are segments of DNA that are especially prone to genetic alteration. The genome is composed of one to several long molecules of DNA, and mutation can occur potentially anywhere on these molecules at any time. The most serious changes take place in the functional units of DNA, the genes. A mutated form of a gene is called a mutant allele. A gene is typically composed of a regulatory region, which is responsible for turning the gene's transcription on and off at the appropriate times during development, and a coding region, which carries the genetic code for the structure of a functional molecule, generally a protein. A protein is a chain of usually several hundred amino acids. Cells make 20 common amino acids, and it is the unique number and sequence of these that give a protein its specific function. Each amino acid is encoded by a unique sequence, or codon, of three of the four possible base pairs in the DNA (A-T, T-A, G-C, and C-G, the individual letters referring to the four nitrogenous bases adenine, thymine, guanine, and cytosine). Mutations are of several types. Changes within genes are called point mutations. The simplest kinds are changes to single base pairs, called base-pair substitutions. Many of these substitutes an incorrect amino acid in the corresponding position in the encoded protein, and of these a large proportion result in altered protein function. Some base-pair substitutions produce a stop codon.

CONCLUSION

At the level of whole populations of organisms, mutation can be viewed as a constantly dripping faucet introducing mutant alleles into the population, a concept described as mutational pressure. The rate of mutation differs for different genes and organisms. In RNA viruses, such as the human immunodeficiency virus (HIV; see AIDS), replication of the genome takes place within the host cell using a mechanism that is prone to error. Hence, mutation rates in such viruses are high. In general, however, the fate of individual mutant alleles is never certain. Most are eliminated by chance. In some cases a mutant allele can increase in frequency by chance, and then individuals expressing the allele can be subject to selection, either positive or negative. Hence, for any one gene the frequency of a mutant allele in a population is determined by a combination of mutational pressure, selection, and chance.