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Section 12 4 mutations answers

Thank you for your participation! The development and function of an organism is in large part controlled by genes. Mutations can lead to changes in the structure of an encoded protein or to a decrease or complete loss in its expression. Because a change in the DNA sequence affects all copies of the encoded protein, mutations can be particularly damaging to a cell or organism. In contrast, any alterations in the sequences of RNA or protein molecules that occur during their synthesis are less serious because many copies of each RNA and protein are synthesized.Geneticists often distinguish between the genotype and phenotype of an organism. Strictly speaking, the entire set of genes carried by an individual is its genotype, whereas the function and physical appearance of an individual is referred to as its phenotype. However, the two terms commonly are used in a more restricted sense: genotype usually denotes whether an individual carries mutations in a single gene (or a small number of genes), and phenotype denotes the physical and functional consequences of that genotype.A fundamental genetic difference between organisms is whether their cells carry a single set of chromosomes or two copies of each chromosome. The former are referred to as haploid; the latter, as diploid. Many simple unicellular organisms are haploid, whereas complex multicellular organisms (e.g., fruit flies, mice, humans) are diploid.Different forms of a gene (e.g., normal and mutant) are referred to as alleles. Since diploid organisms carry two copies of each gene, they may carry identical alleles, that is, be homozygous for a gene, or carry different alleles, that is, be heterozygous for a gene. A recessive mutation is one in which both alleles must be mutant in order for the mutant phenotype to be observed; that is, the individual must be homozygous for the mutant allele to show the mutant phenotype. In contrast, the phenotypic consequences of a dominant mutation are observed in a heterozygous individual carrying one mutant and one normal allele (Figure 8-1).Recessive mutations inactivate the affected gene and lead to a loss of function. For instance, recessive mutations may remove part of or all the gene from the chromosome, disrupt expression of the gene, or alter the structure of the encoded protein, thereby altering its function. Conversely, dominant mutations often lead to a gain of function. For example, dominant mutations may increase the activity of a given gene product, confer a new activity on the gene product, or lead to its inappropriate spatial and temporal expression. Dominant mutations, however, may be associated with a loss of function. In some cases, two copies of a gene are required for normal function, so that removing a single copy leads to mutant phenotype. Such genes are referred to as haplo-insufficient. In other cases, mutations in one allele may lead to a structural change in the protein that interferes with the function of the wild-type protein encoded by the other allele. These are referred to as dominant negative mutations.Some alleles can be associated with both a recessive and a dominant phenotype. For instance, fruit flies heterozygous for the mutant Stubble (Sb) allele have short and stubby body hairs rather than the normal long, slender hairs; the mutant allele is dominant in this case. In contrast, flies homozygous for this allele die during development. Thus the recessive phenotype associated with this allele is lethal, whereas the dominant phenotype is not.Recessive and dominant mutations can be distinguished because they exhibit different patterns of inheritance. To understand why, we need to review the type of cell division that gives rise to gametes (sperm and egg cells in higher plants and animals). The body (somatic) cells of most multicellular organisms divide by mitosis (see Figure 1-10), whereas the germ cells that give rise to gametes undergo meiosis. Like body cells, premeiotic germ cells are diploid, containing two of each morphologic type of chromosome. Because the two members of each such pair of homologous chromosomes are descended from different parents, their genes are similar but not usually identical. Single-celled organisms (e.g., the yeast S. cerevisiae) that are diploid at some phase of their life cycle also undergo meiosis (see Figure 10-54).Figure 8-2 depicts the major events in meiosis. One round of DNA replication, which makes the cell 4n, is followed by two separate cell divisions, yielding four haploid (1n) cells that contain only one chromosome of each homologous pair. The apportionment, or segregation, of homologous chromosomes to daughter cells during the first meiotic division is random; that is, the maternally and paternally derived members of each pair, called homologs, segregate independently, yielding germ cells with different mixes of paternal and maternal chromosomes. Thus parental characteristics are reassorted randomly into each new germ cell during meiosis. The number of possible varieties of meiotic segregants is 2n, where n is the haploid number of chromosomes. In the case of a single chromosome, as illustrated in Figure 8-2, meiosis gives rise to two types of gametes; one type carries the maternal homolog and the other carries the paternal homolog.Now, let's see what phenotypes are generated by mating of wild-type individuals with mutants carrying either a dominant or a recessive mutation. As shown in Figure 8-3a, half the gametes from an individual heterozygous for a dominant mutation in a particular gene will have the wild-type allele, and half will have the mutant allele. Since fertilization of female gametes by male gametes occurs randomly, half the first filial (F1) progeny resulting from the cross between a normal wild-type individual and a mutant individual carrying a single dominant allele will exhibit the mu-tant phenotype. In contrast, all the gametes produced by a mutant homozygous for a recessive mutation will carry the mutant allele. Thus, in a cross between a normal individual and one who is homozygous for a recessive mutation, none of the F1 progeny will exhibit the mutant phenotype (Figure 8-3b). However, one-fourth of the progeny from parents both heterozygous for a recessive mutation will show the mutant phenotype.A mutation involving a change in a single base pair, often called a point mutation, or a deletion of a few base pairs generally affects the function of a single gene (Figure 8-4a). Changes in a single base pair may produce one of three types of mutation:Small deletions have effects similar to those of frameshift mutations, although one third of these will be in-frame and result in removal of a small number of contiguous amino acids.The second major type of mutation involves large-scale changes in chromosome structure and can affect the functioning of numerous genes, resulting in major phenotypic consequences. Such chromosomal mutations (or abnormalities) can involve deletion or insertion of several contiguous genes, inversion of genes on a chromosome, or the exchange of large segments of DNA between nonhomologous chromosomes (Figure 8-4b).Mutations arise spontaneously at low frequency owing to the chemical instability of purine and pyrimidine bases and to errors during DNA replication. Natural exposure of an organism to certain environmental factors, such as ultraviolet light and chemical carcinogens (e.g., aflatoxin B1), also can cause mutations.A common cause of spontaneous point mutations is the deamination of cytosine to uracil in the DNA double helix. Subsequent replication leads to a mutant daughter cell in which a T·A base pair replaces the wild-type C·G base pair. Another cause of spontaneous mutations is copying errors during DNA replication. Although replication generally is carried out with high fidelity, errors occasionally occur. Figure 8-5 illustrates how one type of copying error can produce a mutation. In the example shown, the mutant DNA contains nine additional base pairs.In order to increase the frequency of mutation in experimental organisms, researchers often treat them with high doses of chemical mutagens or expose them to ionizing radiation. Mutations arising in response to such treatments are referred to as induced mutations. Generally, chemical mutagens induce point mutations, whereas ionizing radiation gives rise to large chromosomal abnormalities.Ethylmethane sulfonate (EMS), a commonly used mutagen, alkylates guanine in DNA, forming O6-ethylguanine (Figure 8-6a). During subsequent DNA replication, O6-ethylguanine directs incorporation of deoxythymidylate, not deoxycytidylate, resulting in formation of mutant cells in which a G·C base pair is replaced with an A·T base pair (Figure 8-6b). The causes of mutations and the mechanisms cells have for repairing alterations in DNA are discussed further in Chapter 12.Many common human diseases, often devastating in their effects, are due to mutations in single genes. Genetic diseases arise by spontaneous mutations in germ cells (egg and sperm), which are transmitted to future generations. For example, sickle-cell anemia, which affects 1 in 500 individuals of African descent, is caused by a single missense mutation at codon 6 of the β-globin gene; as a result of this mutation, the glutamic acid at position 6 in the normal protein is changed to a valine in the mutant protein. This alteration has a profound effect on hemoglobin, the oxygen-carrier protein of erythrocytes, which consists of two α-globin and two β-globin subunits (see Figure 3-11). The deoxygenated form of the mutant protein is insoluble in erythrocytes and forms crystalline arrays. The erythrocytes of affected individuals become rigid and their transit through capillaries is blocked, causing severe pain and tissue damage. Because the erythrocytes of heterozygous individuals are resistant to the parasite causing malaria, which is endemic in Africa, the mutant allele has been maintained. It is not that individuals of African descent are more likely than others to acquire a mutation causing the sickle-cell defect, but rather the mutation has been maintained in this population by interbreeding.Spontaneous mutation in somatic cells (i.e., non-germline body cells) also is an important mechanism in certain human diseases, including retinoblastoma, which is associated with retinal tumors in children (see Figure 24-11). The hereditary form of retinoblastoma, for example, results from a germ-line mutation in one Rb allele and a second somatically occurring mutation in the other Rb allele (Figure 8-7a). When an Rb heterozygous retinal cell undergoes somatic mutation, it is left with no normal allele; as a result, the cell proliferates in an uncontrolled manner, giving rise to a retinal tumor. A second form of this disease, called sporadic retinoblastoma, results from two independent mutations disrupting both Rb alleles (Figure 8-7b). Since only one somatic mutation is required for tumor development in children with hereditary retinoblastoma, it occurs at a much higher frequency than the sporadic form, which requires acquisition of two independently occurring somatic mutations. The Rb protein has been shown to play a critical role in controlling cell division (Chapter 13).In a later section, we will see how normal copies of disease-related genes can be isolated and cloned.SUMMARY Diploid organisms carry two copies (alleles) of each gene, whereas haploid organisms carry only one copy. Mutations are alterations in DNA sequences that result in changes in the structure of a gene. Both small and large DNA alterations can occur spontaneously. Treatment with ionizing radiation or various chemical agents increases the frequency of mutations. Recessive mutations lead to a loss of function, which is masked if a normal copy of the gene is present. For the mutant phenotype to occur, both alleles must carry the mutation. Dominant mutations lead to a mutant phenotype in the presence of a normal copy of the gene. The phenotypes associated with dominant mutations may represent either a loss or a gain of function. In meiosis, a diploid cell undergoes one DNA replication and two cell divisions, yielding four haploid cells (Figure 8-2). The members of each pair of homologous chromosomes segregate independently during meiosis, leading to the random reassortment of maternal and paternal alleles in the gametes.

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