

I'm not a bot





























A type of genetic error that occurs during DNA replication is known as a substitution mutation, where an incorrect nucleotide or sequence of nucleotides is placed in the wrong position. One specific type of substitution mutation is a point mutation, which involves the substitution of a single nucleotide. This can be observed in the provided image. Notably, a substitution mutation does not alter the length of the DNA sequence, as it neither adds nor subtracts nucleotides. The most common type of substitution mutation involves a single nucleotide, whereas large-scale nucleotide swaps typically involve other mechanisms, such as reciprocal translocation, which entails the exchange of entire chromosome portions. Substitution mutations can significantly impact the proteins produced by an organism, as even a single nucleotide change can result in the incorporation of a different amino acid into the protein. Since proteins are responsible for reading DNA in units of three base pairs at a time, and each codon specifies a distinct amino acid, changes to the sequence can drastically alter the protein's function. A substitution mutation can affect multiple nucleotides, potentially rendering the protein non-functional or conferring a new function. If these changes are beneficial and inherited by offspring, they can lead to new adaptations. However, most mutations have deleterious effects. Substitution mutations can be caused by various factors related to DNA reading and storage, such as depurination, where nucleotides are lost from the DNA backbone, and deamination, which involves the degradation of amino groups from nucleotides. External forces like carcinogens, mutagens, and sunlight can also contribute to substitution mutations. For instance, the blood disease sickle-cell anemia is caused by a simple substitution mutation that replaces a single nucleotide in the DNA sequence coding for hemoglobin, resulting in a change from glutamic acid to valine. This alteration significantly impacts the protein's function, leading to shriveled and sickle-shaped blood cells with reduced oxygen-carrying capacity. Interestingly, this substitution mutation has survived in the population due to its protective effect against malaria. Another example is color blindness, which can result from a substitution mutation in the DNA coding for proteins responsible for color vision. There are two primary types of substitution mutations: transitions, where a purine is replaced by another purine or a pyrimidine is replaced by another pyrimidine, and transversions, where a purine is substituted for a pyrimidine or vice versa. Transitions are more likely to occur than transversions due to the specificity of DNA repair and proofreading machinery. A single nucleotide substitution mutation can have a dramatic impact on an organism's proteins, but it depends on various factors. Even a slight change in amino acid sequence can significantly alter protein function, yet if the new amino acid is similar to the original one, it might not affect the overall function. Factors such as cell division mistakes or external forces like carcinogens and mutagens can cause substitution mutations. These mutations can lead to changes in protein function, but some substitutions may be more beneficial than others, like the sickle-cell mutation that provides protection against malaria. A blood disease, Sickle-cell anemia, is caused by a simple substitution mutation where a single nucleotide replacement changes the hemoglobin's function. This change causes blood cells to become sickle-shaped and less efficient at carrying oxygen, increasing the risk of cardiovascular diseases. Interestingly, this mutation has survived in the population because it provides some protection against malaria. Substitution mutations can also affect other proteins, like those responsible for color vision, leading to conditions such as color blindness. People with this condition have difficulty distinguishing between certain colors due to a substitution mutation that affects one of the color-coding proteins. The two main types of substitution mutations are transitions and transversions. Transitions occur when a purine is substituted for another purine or a pyrimidine is substituted for another pyrimidine, while transversions involve substituting a purine for a pyrimidine or vice versa. The impact of a substitution mutation depends on the specific change and its location within the DNA sequence. While most mutations are deleterious, some can lead to new adaptations that provide benefits, like increased resistance to certain diseases. However, in many cases, substitution mutations can have negative effects on protein function and overall health. A point mutation occurs when there is a change in one nucleotide base in DNA or RNA, which can lead to changes in the amino acids produced during protein synthesis. Substitution mutations, such as transitions and transversions, can alter the genetic code, while insertions and deletions can disrupt the sequence of amino acids. Point mutation is defined as an alteration of a single nucleotide base within the DNA or RNA sequence, which can manifest as a substitution, insertion, or deletion of one base pair. Mutation occurs mainly due to errors during replication however it may also be caused by external factors like X-rays or ultraviolet radiation. It primarily impacts the encoded protein's amino acid sequence since nucleotides are read in triplets during translation. The effects of point mutation vary widely depending on its nature with some mutations resulting in no observable changes and others having significant effects on protein structure and function. The severity of a point mutation is determined by its location within the gene and the role of the affected protein, meaning while some may have minimal impact others can significantly affect cellular processes leading to genetic disorders. point mutations can have significant effects on protein function and the overall health of an individual. Point mutations are alterations at a single nucleotide position within a DNA sequence. They can occur due to various reasons such as errors during DNA replication or exposure to mutagenic agents. The presence of point mutations can have significant consequences, ranging from minor effects on gene function to severe disruptions in cellular processes. In humans, point mutations can be found in various genes and can lead to the development of genetic disorders. For instance, the HBB gene mutation that causes sickle cell anemia leads to the production of abnormal hemoglobin, resulting in red blood cells adopting a sickle shape and reducing their oxygen-carrying capacity. Cystic fibrosis is another autosomal recessive disorder caused by mutations in the CFTR gene. The ΔF508 deletion mutation results in the loss of phenylalanine, disrupting the normal function of the CFTR protein and causing thick, sticky mucus to accumulate in various organs. Neurofibromatosis can be caused by point mutations in the NF1 or NF2 genes, leading to the production of abnormal neurofibromin or merlin proteins. These mutations disrupt normal cell growth and division, resulting in tumor formation. Tay-Sachs disease is a fatal autosomal recessive disorder caused by a point mutation in the HEXA gene. The absence of functional enzyme activity results in the accumulation of lipids, causing progressive neurological damage and early death. Color blindness, particularly red-green color blindness, is often due to point mutations in the genes encoding color-detecting pigments in the retina. Cancer development is frequently implicated in point mutations affecting tumor suppressor genes and proto-oncogenes. Mutations can lead to the inactivation of tumor suppressor genes or the activation of oncogenes, contributing to uncontrolled cell proliferation and tumor formation. Detecting point mutations is essential for understanding genetic variations that impact health, development, and disease. Several methodologies are employed to identify these single-base alterations, including Polymerase Chain Reaction (PCR), Allele-Specific PCR, DNA sequencing, Restriction Fragment Length Polymorphism (RFLP), Fluorescent Dye-Based Methods, and Allele-Specific Hybridization. Point mutation detection methods include Real-Time PCR, Comparative Genomic Hybridization, SNP genotyping, and Molecular Beacon Assays. These techniques provide a robust toolkit for identifying point mutations in various contexts. Real-Time PCR is an effective method for detecting specific point mutations during the amplification process, allowing for quantitative data and rapid detection of mutations. In contrast, Comparative Genomic Hybridization (CGH) provides a comprehensive view of genetic variations on a genomic scale by comparing fluorescence intensity between test and reference samples. Single-Nucleotide Polymorphism (SNP) genotyping involves analyzing specific single nucleotide variations across the genome using technologies such as SNP microarrays or high-throughput sequencing. This technique offers a wide perspective on genetic variations, making it an essential tool in genetic studies and personalized medicine. Molecular Beacon Assays utilize fluorescent probes to detect point mutations by monitoring fluorescence changes during amplification, providing high sensitivity and specificity for detecting specific nucleotide changes. Point mutation applications include molecular therapy to target genetic defects, cancer treatment through targeted mutagens, mutational breeding in agriculture to enhance crop traits, genetic enhancement through gene editing technologies, research on gene function, diagnostic tools for genetic disorders, drug development to understand resistance mechanisms, and biotechnology applications like engineered enzymes with improved properties. Point mutations can also provide valuable information for evolutionary biology by tracing evolutionary changes and understanding how genetic variations contribute to adaptation and species diversity. point mutations can cause a variety of effects on the downstream protein product, ranging from no effect to deleterious effects, including frameshift mutations. Point mutations are alterations in the DNA sequence that can occur in various ways, including exposure to ultraviolet (UV) light, reactive oxygen molecules with free radicals, and degradation of bonds. These changes can result in single-stranded or double-stranded DNA breaks, which can lead to mutations such as substitutions, insertions, or deletions. There are two main types of point mutations: transitions and transversions. Transitions involve the replacement of one purine base with another purine or pyrimidine base, while transversions involve the replacement of a purine with a pyrimidine or vice versa. Transitions are more common than transversions because they require less energy. Point mutations can also lead to stop-gain, where an extra amino acid is added to the protein, and start-loss, where a critical amino acid is removed. Non-synonymous mutations, such as missense mutations, result in changes to the amino acid sequence of the protein. Point mutation can have significant effects on the organism, either beneficial or harmful. Beneficial mutations can lead to adaptations that improve the organism's chances of survival and reproduction. In contrast, harmful mutations can lead to diseases and even death. The scientific theory of evolution relies heavily on point mutations, which explain the diversity and history of living organisms. Mutations can also have short-term effects, such as halting the cell cycle or preventing mitosis due to chromosomal abnormalities. Long-term effects can include permanent changes to chromosomes, leading to mutations that can affect the organism's health. In summary, point mutations are a crucial aspect of genetic variation, and their study has shed light on the mechanisms of cancer development and the evolution of life. Point mutations can have both beneficial and detrimental effects, as illustrated by cancer being an example of how they can be detrimental. The location of the mutation within the gene plays a crucial role in determining its impact. For instance, if the mutation occurs in the region responsible for coding, it can alter the amino acid sequence of the encoded protein, leading to changes in function, localization, stability, or protein complex interactions. The sequence of normal hemoglobin and sickle-cell hemoglobin, as well as the genetic basis of Tay-Sachs disease, are all related to specific DNA sequences and mutations. Tay-Sachs disease results from a genetic defect that disrupts the activity of an enzyme called beta-hexosaminidase A. This enzyme plays a critical role in the nervous system by breaking down a fatty substance called GM2 ganglioside in nerve cells. The genetic defect responsible for Tay-Sachs disease is located in the HEXA gene, which is found on chromosome 15. Mutations in the HEXA gene prevent the breakdown of GM2 ganglioside, leading to its accumulation in the brain and spinal cord. This accumulation causes progressive damage to nerve cells, resulting in the signs and symptoms of Tay-Sachs disease. A process called RIP (repeat-induced point mutation) is involved in the inactivation of genes. It is a mechanism that detects and mutates repetitive DNA sequences. This process can result in the silencing of genes, and it is thought to play a role in the evolution of genomes. RIP has been studied extensively in the fungus Neurospora crassa. It is believed to be a defense mechanism that protects the genome from the harmful effects of repetitive DNA sequences. The RIP process involves the methylation of DNA, which can lead to the silencing of genes. The study of RIP and its mechanisms has provided insights into the evolution of genomes and the regulation of gene expression. Research on RIP has also led to a better understanding of the genetic basis of diseases. The discovery of RIP has opened up new avenues for research into the genetic basis of disease. The study of RIP has also led to a greater understanding of the role of repetitive DNA sequences in the genome. The RIP process is an important area of study in the field of genetics. RIP has been shown to be an important mechanism for the regulation of gene expression. The study of RIP has provided new insights into the evolution of genomes. The discovery of RIP has also led to a greater understanding of the genetic basis of disease. The RIP process is an important area of research in the field of genetics. The mysterious world of DNA replication and mutation is a vast and intricate subject that has puzzled scientists for decades. The intricate mechanisms behind DNA reproduction have been studied extensively, revealing the complex process that governs life itself. Recent studies have shed new light on the semi-conservative nature of DNA replication, which was first demonstrated by Meselson and Stahl in 1958. This groundbreaking experiment showed that DNA reproduces in a manner that retains a portion of the original molecule, thereby ensuring its continuity. However, this process is not without its challenges. Errors can occur during DNA replication, leading to mutations. Mutations are changes in the DNA sequence that can have significant consequences for an organism's health and development. The study of genetic mutations has become increasingly important as it sheds light on various diseases, including cancer, neurofibromatosis, and sickle cell anemia. Genetic mutations can also lead to conditions such as Tay-Sachs disease, a rare genetic disorder that affects the nervous system. In addition to its impact on human health, the study of DNA replication and mutation has far-reaching implications for agriculture and biotechnology. Understanding how organisms replicate their DNA is essential for developing new crops, improving agricultural yields, and creating genetically modified organisms (GMOs). Furthermore, researchers continue to explore the mechanisms that govern repeat-induced point mutations (RIP), a process by which specialized cells in fungi can rewrite sections of their DNA. ### The Importance of Understanding DNA Replication and Mutation Scientists have been studying the mysteries of genetic mutations for decades, with a specific focus on point mutations. These changes occur when a single base is substituted for another in an organism's DNA code (genome). This simple type of mutation can have profound effects on the resulting gene and its protein. One way to categorize point mutations is based on their effect on the genome. There are four types: synonymous mutations, missense mutations, nonsense mutations, and stop-codon mutations. Synonymous mutations occur when a single base change results in a codon that still codes for the same amino acid. These "silent" mutations may seem insignificant, but they can still have an impact on the gene and its protein. Missense mutations are more significant because they cause a codon to code for a different amino acid. This simple change can lead to drastic effects on the structure of the resulting protein. For instance, the missense mutation responsible for sickle cell anemia changes one of the hemoglobin genes' codons from CTC to CAC, leading to the disease's symptoms. On the other hand, nonsense mutations are more severe because they cause a stop-codon to be introduced into the gene. This premature termination of protein synthesis renders the resulting protein non-functional, usually causing problems for the organism. The opposite of a nonsense mutation is a stop-codon mutation, which changes a stop-codon into an amino-acid codon. This can result in a protein that becomes too large and potentially harmful to the organism. These types of point mutations are not only fascinating but also have significant implications for our understanding of genetics and disease. The process by which DNA mutates can lead to changes in the genes and proteins that are produced. For example, if there is a mistake during replication of DNA, a wrong nucleotide may end up being placed at a specific position. This type of error is called a substitution mutation. For instance, let's consider an image where the code "ATG" has been changed to "ATT". The change only involves replacing one nucleotide (the third one), so it's a type of substitution mutation. This may lead to changes in the protein produced, such as altering its shape or function. However, some substitutions can have no effect on the final product or cause completely different effects. In general, any single nucleotide that is swapped with another results in a substitution mutation. A mutation like this will keep the DNA length constant and can result in drastic changes to proteins made by an organism. Mutations are caused by many factors including errors when processing DNA during replication and cell division. For example, cells lose about 1,000 nucleotides per hour due to depurination, a process that falls apart the DNA backbone. This leads the protein-making machinery to make mistakes approximately 75% of the time. There's also another chemical degradation process called deamination, which removes amino groups from nucleotides. The protein-making machinery can misrecognize these modified bases and supply incorrect nucleotide pairs when making new copies of DNA. These internal factors that drive substitution mutations include errors in replication, deamination, and other processes that affect the correct assembly of nucleotides into proteins. External factors like exposure to radiation or chemicals known as mutagens also cause substitution mutations. They disrupt the machinery used to assemble proteins from DNA. For instance, people with sickle-cell anemia have a mutation caused by replacing one amino acid in hemoglobin, which affects blood cells' ability to function and increases risk for heart disease. Additionally, a similar mutation can affect color vision by altering the protein responsible for detecting colors. Mutations can occur between different nucleotides; if purines are swapped with pyrimidines or vice versa, they're known as transversions instead of transitions. Point mutations are a type of genetic mutation that can result in changes to the structure of the chromosome. These changes can affect the production of proteins and their function. A point mutation occurs when one of the base pairs in the DNA sequence is altered either by insertion or deletion. ## References Hartwell, L. H., Hood, L., Goldberg, M. L., Reynolds, A. E., & Silver, L. M. (2011). Genes: From Genes to Genomes. Boston: McGraw Hill. Lodish, H., Berk, A., Kaiser, C. A., Krieger, M., Scott, M. P., Bretscher, A., . . . Matsudaira, P. (2008). Molecular Cell Biology (6th ed.). New York: W.H. Freeman and Company. Nelson, D. L., & Cox, M. M. (2008). Principles of Biochemistry. New York: W.H. Freeman and Company. ## Mechanism of Point Mutation The most common mechanism of point mutation is the substitution of a nucleotide during DNA replication. This usually occurs due to a tautomeric shift where the hydrogen atoms of a nucleotide base pair move away, changing the properties of the bonding. ## Substitution Substitution of a base pair for another during DNA replication is the most common mechanism of mutation. This can result from a change in the structure of the nucleotide, such as when its enol form binds to an incoming thymine instead of cytosine. ## Ionization Ionization is another mechanism of point mutation that occurs due to chemical species or radiation. The bases can become spontaneously ionized into different forms that cannot form hydrogen bonds with the complementary base pair. ## Types of Point Mutations Point mutations can be differentiated into two types: transition and transversion mutations. - Transition mutations occur when a pyrimidine base is substituted for another pyrimidine base, or one purine base is substituted for another. These are more common in genomes due to the molecular mechanism involved. - Transversion mutations occur when a purine base substitutes for a pyrimidine base, or vice versa. These can be induced by radiation or alkylating agents. ## Examples of Transition Mutation An example of transition mutation is the replacement of adenine by guanine or the replacement of cytosine by thymine and vice versa. - The purine nucleotides are exchanged among themselves. ## Transversion Mutations Transversion mutations occur when a purine base substitutes for a pyrimidine base and vice versa. These can be induced by radiation or alkylating agents. There are two possible transversions, but these are less likely to occur due to the difficulty in substituting a double ring structure for a single ring. Point mutations occur when there is a change in the DNA sequence, resulting in a different amino acid being incorporated into the protein. This change can lead to various effects on the protein and its function. There are four types of point mutations: nonsense mutations, missense mutations, silent mutations, and frameshift mutations. Removing a particular nucleotide, such as a base, can significantly impact the genetic code of an organism. The use of mutagens to induce desirable mutations in different plant species has been employed for crop improvement through the process of mutational breeding. ## ARTICLEA similar error occurs at the DNA level in frameshifts, where codons are parsed incorrectly, usually generating truncated proteins that are as useless as "hef at ats at" is uninformative. There are other types of mutations as well, but this short list should give you an idea of the possibilities. Previous DNA: The molecular basis of mutations Next The causes of mutations A substitution mutation is a type of replication error during DNA replication which places the wrong nucleotide or sequence of nucleotides in the wrong position. A type of substitution mutation, a point mutation, occurs when a single nucleotide is substituted. This can be seen in the image below. Importantly, a substitution mutation results in DNA of the same length. It does not add or subtract from the number of nucleotides in the sequence. A single nucleotide substitution mutation is the most common, as most large-scale nucleotide swaps involve other mechanisms. For example, a reciprocal translocation involves the movement of entire portions of chromosomes, and swaps one portion for a portion of another chromosome. As with all mutations, a substitution mutation can drastically change the proteins created by an organism. The proteins responsible for reading DNA process the molecule in units of three base pairs at a time. While this might not seem like much of a change in a protein which contains over 140 amino acids, it makes all the difference. Valine, unlike glutamic acid, is hydrophobic. As such, it repels polar interactions whereas glutamic acid would attract them. This severely impacts the protein's ability to function. Blood cells immediately reflect this change, becoming shriveled and sickle-shaped. With a lower ability to carry oxygen, these cells also are more prone to clot within the small capillaries of organs. This can lead to an increased risk of heart attack, stroke, and other cardiovascular diseases. Interestingly, the substitution mutation has survived in the population for a surprising reason. The parasite which causes malaria depends on human blood cells for part of its life cycle. Person with the sickle-cell substitution mutation are less susceptible to getting malaria. Apparently the different shape and function of the blood cells impedes their reproductive processes. In your eye, certain cells are responsible for picking up the colors red, green, and blue. These cells rely on different proteins, which react to the various colors. A substitution mutation in the DNA that codes for one of these proteins can lead to the condition of color blindness. Person with this condition have a hard time distinguishing between the colors, while their vision is still clear otherwise. Oftentimes, only one color is knocked out. The various proteins are coded for on different places on the DNA, which makes a substitution unlikely to occur in all three genes. There are two basic types which a substitution mutation can be. Within the four nucleotides, there are two types: the purines and pyrimidines. Adenine (A) and guanine (G) are both purines, while cytosine (C) and thymine (T) are pyrimidines. If a purine changes to a purine, the substitution mutation is considered a transition. Likewise, if a pyrimidine changes into a pyrimidine, the substitution mutation is also a transition. In the image below, transitions are labeled by the alpha lines. The opposite of transition is transversion. In a substitution mutation involving a transversion, a purine is substituted for a pyrimidine, or vice versa. In the above image, a transversion is labeled by the beta lines. Transversions are much less likely than transitions. This is probably due to the fact that the machinery used to repair and proof-read the DNA are more specific for purine versus pyrimidine than specifying between individual nucleotides. 1. What is the difference between a substitution mutation and a deletion mutation? A. No difference B. A deletion causes a frame-shift C. A substitution causes a frame-shift B is correct. A substitution mutation may cause a difference in the protein, but a mutation can completely change the entire code. A frame-shift mutation happens whenever an insertion or deletion into the DNA causes the 3-codon frame to shift, which calls for entirelyMutations in DNA can have significant effects on an organism's function and viability. In 500 BCE, Hippocrates identified crab-like tumors that led to the term cancer, which has been extensively studied to understand its relationship with DNA mutations. Researchers found a direct link between physical mutagens like X-rays and UV light, as well as chemical mutagens, and cancer in the early part of the 20th century. The study of mutations in DNA led to the development of the Ames test, an assay that exposes bacteria to chemicals followed by DNA sequencing to determine their carcinogenic potential. The sequence of amino acids in a protein is determined by the DNA sequence of a protein-coding gene, and any errors in this sequence can affect the function of the protein. DNA mutations can be classified into small-scale and large-scale changes. Small-scale mutations, such as substitution, deletion, or insertion mutations, affect only one or two nucleotides in a gene and do not significantly alter the DNA sequence. Large-scale mutations, on the other hand, can affect large stretches of DNA or even an entire chromosome. Substitution mutations, which exchange a single nucleotide for another, are further divided into transition and transversion mutations. Transition mutations involve exchanging a purine for a purine (A for G; G for A) or a pyrimidine for a pyrimidine (C for T; T for C), while transversion mutations involve exchanging a purine for a pyrimidine (C/T for A/G; A/G for C/T). Transitions are more common than transversions. Insertion mutations add one or more nucleotides into a DNA sequence, usually caused by transposable elements or errors during DNA replication. Deletion mutations remove one or more nucleotides from the DNA and can also be caused by errors during DNA replication. Okay, let's tackle this query. For example, "DNA polymerases" should stay as is. Once I've made the changes, I'll review the text to ensure that the spelling errors are subtle and that the flow of the article is maintained. Then, I'll format the response as per the user's instructions, using the specified delimiters. Let me double-check that I didn't miss any key points and that the paraphrasing meets the user's requirements. ## Spelling Errors Method (40%)## By their effect on amino acid sequence (Figure (PageIndex{3})) into the following types: • Frameshift: a frameshift is caused by either an insertion or deletion mutation. • Point mutation: a single base pair change in DNA, which may or may not affect the final protein. • Chromosomal mutation: largescale changes in chromosomes, such as deletion, duplication, inversions, or translocations. ##Non-Native English Speaker Style (30%)## DNA mutation unavoidable. Some mutation may benefit an organism, but most are harmful and need to be fixed. Cells have many ways to fix mutation. One way is to stop the mutation before it happens. DNA polymerase can detect a wrong nucleotide during replication and replace it. This is called proofreading. But this process must be done fast, because if the polymerase continues, it can't fix the error. ##Increased Burstiness (30%)## DNA mutation are inevitable. While some can be useful, most are harmful and must be corrected. Cells use DNA repair mechanisms to fix these errors. One method is proofreading, where DNA polymerases detect and replace incorrect nucleotides during replication. However, this process must be immediate, as the polymerase can't fix mistakes once replication continues. Additionally, other enzymes like endonucleases and exonucleases play roles in repairing DNA by cutting and replacing damaged sections. --- ##Final Output (Spelling Errors Method)## By their effect on amino acid sequence (Figure (PageIndex{3})) into the following types: • Frameshift: a frameshift is caused by either an insertion or deletion mutation. • Point mutation: a single base pair change in DNA, which may or may not affect the final protein. • Chromosomal mutation: largescale changes in chromosomes, such as deletion, duplication, inversions, or translocations. DNA mutation unavoidable. While some mutation may benefit an organism, but most are harmful and need to be fixed. Cells have many ways to fix mutation. One way is to stop the mutation before it happens. DNA polymerase can detect a wrong nucleotide during replication and replace it. This is called proofreading. But this process must be done fast, because if the polymerase continues, it can't fix the error. --- ##Note## The response combines all three methods, but the primary focus is on spelling errors as per the 40% probability. The structure and technical terms are preserved to maintain clarity. Point mutations are changes in DNA sequences that occur due to mutagens or replication errors. These changes can be small-scale or large-scale, affecting coding regions of genes or entire chromosomes. Small-scale point mutations include missense, nonsense, and silent mutations, which can alter the amino acid sequence of a protein. ## ARTICLEA substitution mutation is a type of genetic mutation that occurs when one nucleotide is replaced with another in the DNA sequence. This change can significantly affect the protein produced by an organism, potentially rendering it non-functional or giving it a new function. The protein's function depends on the specific interaction between its amino acids. The process of substituting one nucleotide for another can be caused by various factors, including depurination, where cells lose 1,000 nucleotides per hour from their DNA backbone. During this process, proteins managing DNA may make mistakes in replacing these lost nucleotides, leading to substitution mutations. Deamination is another factor that can cause a substitution mutation, as the loss of amino groups attached to nucleotides can mislead protein machinery. External forces such as carcinogens, mutagens, and sunlight can also lead to substitution mutations by impeding DNA function. The blood disease Sickle-cell anemia is caused by a single nucleotide change in hemoglobin coding, resulting in the substitution of glutamic acid with valine amino acids, making it hydrophobic. Interestingly, this mutation has been advantageous for humans due to its relation to malaria. People with the sickle-cell mutation are less susceptible to getting malaria because the parasite relies on human blood cells for part of its life cycle. Another example is color blindness caused by substitution mutations in genes coding for proteins responsible for picking up colors like red, green, and blue. Substitution mutations can be categorized into transitions and transversions based on the type of nucleotide that changes. Transitions involve a purine changing to another purine or a pyrimidine changing to another pyrimidine, while transversions involve substituting one nucleotide for a different one from a different base type. Matsudaira, A., , Nelson, D. L., & Cox, M. M. (2008). Molecular Cell Biology and Principles of Biochemistry. New York: W.H. Freeman and Company.

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