How gene mutation works

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DNA mutations happen when there are changes in the nucleotide sequence that makes up the strand of DNA. Image by: BlackJack3D/Getty Images Image: BlackJack3D/Getty Images

Genes are segments of DNA located on chromosomes. A gene mutation is defined as an alteration in the sequence of nucleotides in DNA. This change can affect a single nucleotide pair or larger gene segments of a chromosome. DNA consists of a polymer of nucleotides joined together. During protein synthesis, DNA is transcribed into RNA and then translated to produce proteins. Altering nucleotide sequences most often results in nonfunctioning proteins. Mutations cause changes in the genetic code that lead to genetic variation and the potential to develop the disease. Gene mutations can be generally categorized into two types: point mutations and base-pair insertions or deletions.

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Point Mutations

Point mutations are the most common type of gene mutation. Also called a base-pair substitution, this type of mutation changes a single nucleotide base pair. Point mutations can be categorized into three types:

Silent Mutation: Although a change in the DNA sequence occurs, this type of mutation does not change the protein that is to be produced. This is because multiple genetic codons can encode for the same amino acid. Amino acids are coded for by three-nucleotide sets called codons. For example, the amino acid arginine is coded for by several DNA codons including CGT, CGC, CGA, and CGG (A = adenine, T = thymine, G = guanine, and C = cytosine). If the DNA sequence CGC is changed to CGA, the amino acid arginine will still be produced.

Missense Mutation: This type of mutation alters the nucleotide sequence so that different amino acid is produced. This change alters the resulting protein. The change may not have much effect on the protein, may be beneficial to protein function, or may be dangerous. Using our previous example, if the codon for arginine CGC is changed to GGC, the amino acid glycine will be produced instead of arginine.

Nonsense Mutation: This type of mutation alters the nucleotide sequence so that a stop codon is coded for in place of amino acid. A stop codon signals the end of the translation process and stops protein production. If this process is ended too soon, the amino acid sequence is cut short and the resulting protein is most always nonfunctional.

Base-Pair Insertions And Deletions

Mutations can also occur in which nucleotide base pairs are inserted into or deleted from the original gene sequence. This type of gene mutation is dangerous because it alters the template from which amino acids are read. Insertions and deletions can cause frame-shift mutations when base pairs that are not a multiple of three are added to or deleted from the sequence. Since the nucleotide sequences are read in groupings of three, this will cause a shift in the reading frame. For example, if the original, transcribed DNA sequence is CGA CCA ACG GCG ..., and two base pairs (GA) are inserted between the second and third groupings, the reading frame will be shifted.

Original Sequence: CGA-CCA-ACG-GCG...

Amino Acids Produced: Arginine/Proline/Threonine/Alanine...

Inserted Base Pairs (GA): CGA-CCA-GAA-CGG-CG...

Amino Acids Produced: Arginine/Proline/Glutamic Acid/Arginine...

The insertion shifts the reading frame by two and changes the amino acids that are produced after the insertion. The insertion can code for a stop codon too soon or too late in the translation process. The resulting proteins will be either too short or too long. These proteins are for the most part defunct.

DNA Mutation Analogy

Much like reading text, the DNA sequence is "read" by messenger RNA to produce a "story" or an amino acid chain that will be used to make a protein. Since each codon is three letters long, let's

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see what happens when a "mutation" occurs in a sentence that uses only three letter words.

THE RED CAT ATE THE RAT.

If there was a point mutation, the sentence would change to:

THC RED CAT ATE THE RAT.

The "e" in the word "the" mutated into the letter "c". While the first word in the sentence is no longer the same, the rest of the words still make sense and are what they are supposed to be.

If an insertion were to mutate the above sentence, then it might read:

THE CRE DCA TAT ETH ERA T.

The insertion of the letter "c" after the word "the" completely changes the rest of the sentence. Not only is the second word no longer readable, neither are any words after it. The entire sentence has changed into nonsense.

A deletion would do something similar to the sentence:

THE EDC ATA TET HER AT.

In the example above, the "r" that should have come after the word "the" has been deleted. Again, it changes the entire sentence. Even though in this example, some of the subsequent words are readable, the meaning of the sentence has completely changed. This shows that even if codons are changed into something that isn't nonsense, it still completely changes the protein into something that is no longer functional.

Causes Of Gene Mutation

Gene mutations are most commonly caused as a result of two types of occurrences. Environmental factors such as chemicals, radiation, and ultraviolet light from the sun can cause mutations. These mutagens alter DNA by changing nucleotide bases and can even change the shape of DNA. These changes result in errors in DNA replication and transcription.

Other mutations are caused by errors made during mitosis and meiosis. Common errors that occur during cell division can result in point mutations and frameshift mutations. Mutations during cell division can lead to replication errors which can result in the deletion of genes, translocation of portions of chromosomes, missing chromosomes, and extra copies of chromosomes.

Genetic Disorders

According to the National Human Genome Institute, almost all diseases have some sort of genetic factor. These disorders can be caused by a mutation in a single gene, multiple gene mutations, combined gene mutation, and environmental factors, or by chromosome mutation or damage. Gene mutations have been identified as the cause of several disorders including sickle cell anemia, cystic fibrosis, Tay-Sachs disease, Huntington disease, hemophilia, and some cancers.

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Write Prompt

Read the following claim.

Are the results of a gene mutation always harmful?

What evidence from the article supports this claim? Explain why the evidence supports this claim.

Write Prompt

Choose a process or event from the text. Describe the process or event using a short narrative to highlight the most important information.

Quiz

1 Which of the following MOST influences a major shift in how the entire nucleotide sequence is read?

(A) base-pair substitutions and point mutations

(B) silent mutations and nonfunctioning proteins

(C) missense mutations and nonsense mutations

(D) base-pair insertions and deletions

2 According to the article, why do point mutations and frameshift mutations occur?

(A) They occur as a result of factors such as chemicals, radiation and ultraviolet light.

(B) They occur as a result of errors that happen during mitosis and meiosis.

(C) They occur as a result of mutagens that change nucleotide bases and even the shape of DNA.

(D) They occur as the result of genetic disorders such as sickle cell anemia and cystic fibrosis.

3 What is the main reason the author includes the section "DNA Mutation Analogy"?

(A) to highlight the three types of point mutations and what makes them similar and different

(B) to explain what environmental factors cause genetic mutations to occur and the results of those mutations

(C) to provide a simple way of helping the reader better understand the different types of genetic mutations

(D) to introduce the idea that frameshift mutations are much more dangerous than point mutations

4 Read the introduction [paragraph 1]. How does the introduction introduce the main idea?

(A) by explaining what types of diseases are caused by genetic mutations

(B) by listing and defining the three different types of point mutation

(C) by comparing and contrasting the two most common types of genetic mutations

(D) by defining genetic mutation and explaining its effects