MUTATION 101

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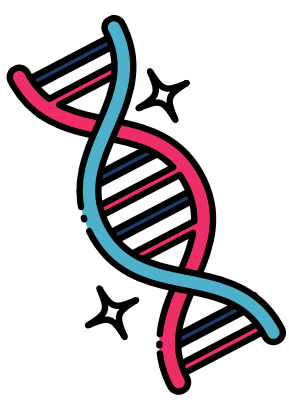
What is Mutation?

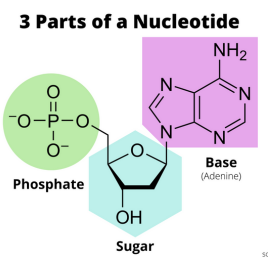
Mutations are changes in the genetic information of a cell or a virus. These changes can affect both the structure and the function of the protein that that DNA sequence transcripts and/or translates to.



Hold up!

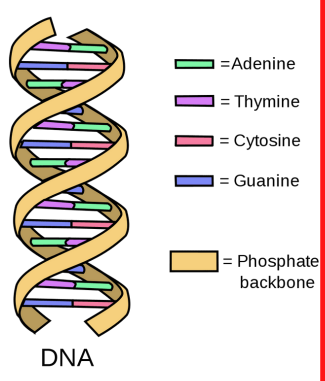
What is a Gene?

A gene is a sequence of DNA or RNA that codes for a protein or trait. Genes can be passed to an organism's offspring, being the basis of inheritance.



What is DNA?

DNA is short for deoxyribonucleic acid, which is a polymer chain made of smaller units -monomers- called nucleotides. There are different kinds of nucleotides that vary on their base.



DNA

Back to mutations!

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What causes a Mutation?

They are spontaneous or caused by environmental factors such as radiation (i.e. gamma, UV) or chemicals.

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Mutation Types

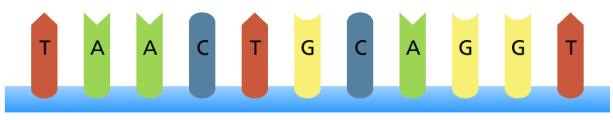
1. Point Mutations
2. Insertion
3. Deletion
4. Substitution
5. Inversion

SOURCES

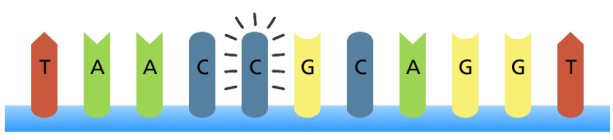
https://medlineplus.gov/genetics/understanding/basics/gene/
https://www.nature.com/scitable/knowledge/library/mutations-are-the-raw-materials-of-evolution-17395346/
https://kids.britannica.com/kids/article/DNA/390730
Pictures:
https://sciencenotes.org/what-are-the-three-parts-of-a-nucleotide/
https://www.yourgenome.org/facts/what-types-of-mutation-are-there

1. Point Mutations

Original sequence

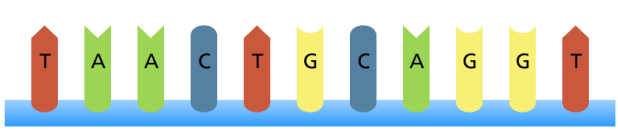


Point mutation

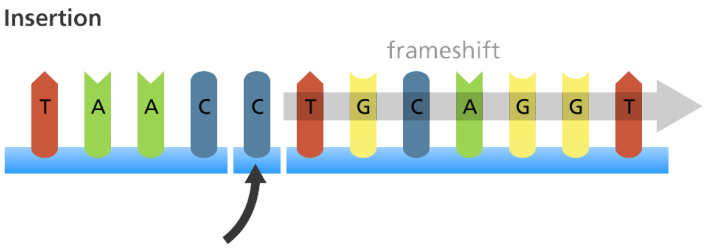


2. Insertion

Original sequence

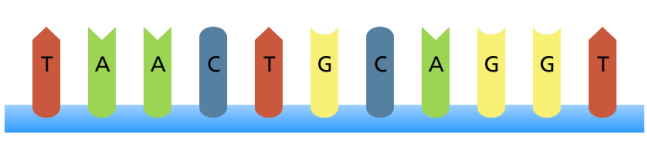


Insertion

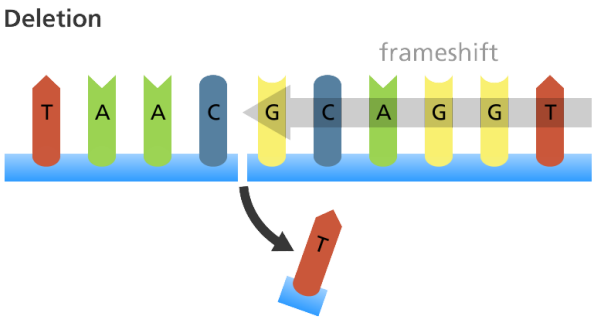


3. Deletion

Original sequence

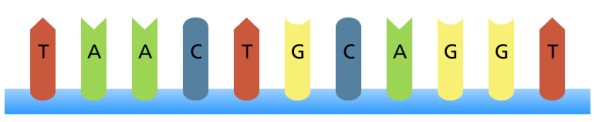


Deletion

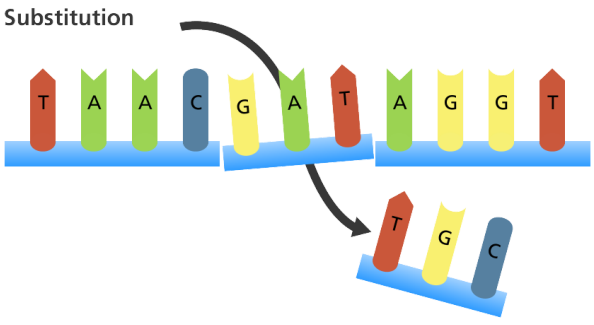


4. Substitution

Original sequence

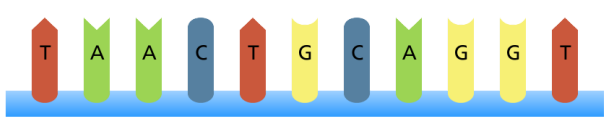


Substitution

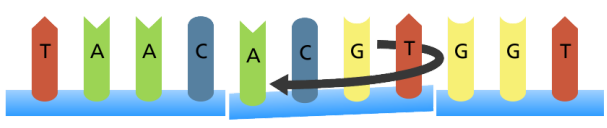


5. Inversion

Original sequence



Inversion



SOURCES

Pictures:
https://www.yourgenome.org/facts/what-types-of-mutation-are-there

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Consequences of Mutations

- **Silent:** same amino acid, the mutation that has no apparent effect on the protein.
- **Missense:** different amino acid, little effect on the protein.
- **Nonsense:** introduce a 'stop' triplet, smaller protein.
- **Frameshift:** the protein that is made is most of the timenonfunctional.

	substitution	addition	deletion
silent	✕		
missense	✕		
nonsense	✕	✕	✕
frame shift		✕	✕

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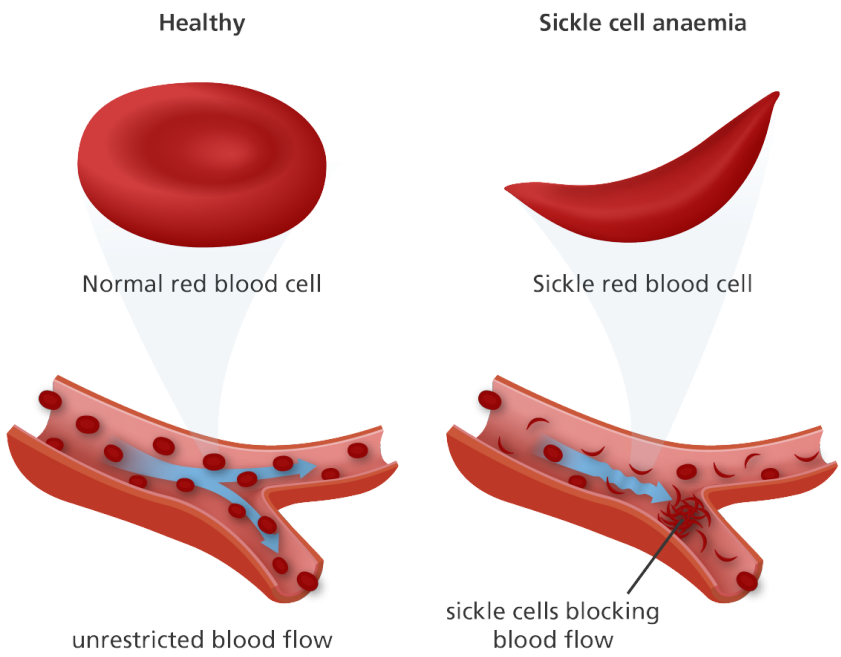
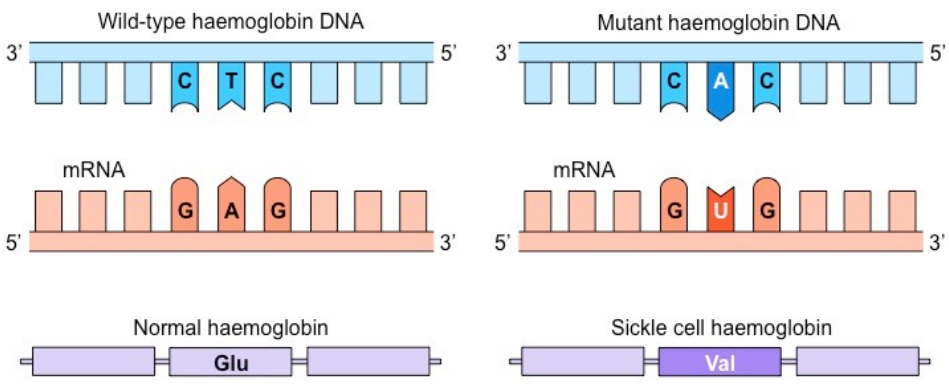
Mutation in Humans: Sickle Cell Anaemia

The gene that causes sickle cell anaemia is different from the wild-type (normal) gene by a single DNA nucleotide.



The mutant (sickle-cell) strand has an A where the wild- type strand has a T. This leads to mutant β-globin, which has a valine (Val) instead of a glutamic acid (Glu).



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Mutation Missconception

Mutations aren't always a bad thing! They are the ultimate source of new genes, which leads to diversity among organisms and change over time - evolution!

SOURCES

http://biology4alevel.blogspot.com/2016/06/133-genetic-mutations.html

Pictures:

https://ib.bioninja.com.au/standard-level/topic-3-genetics/31-genes/mutations.html

https://www.yourgenome.org/facts/what-is-sickle-cell-anaemia