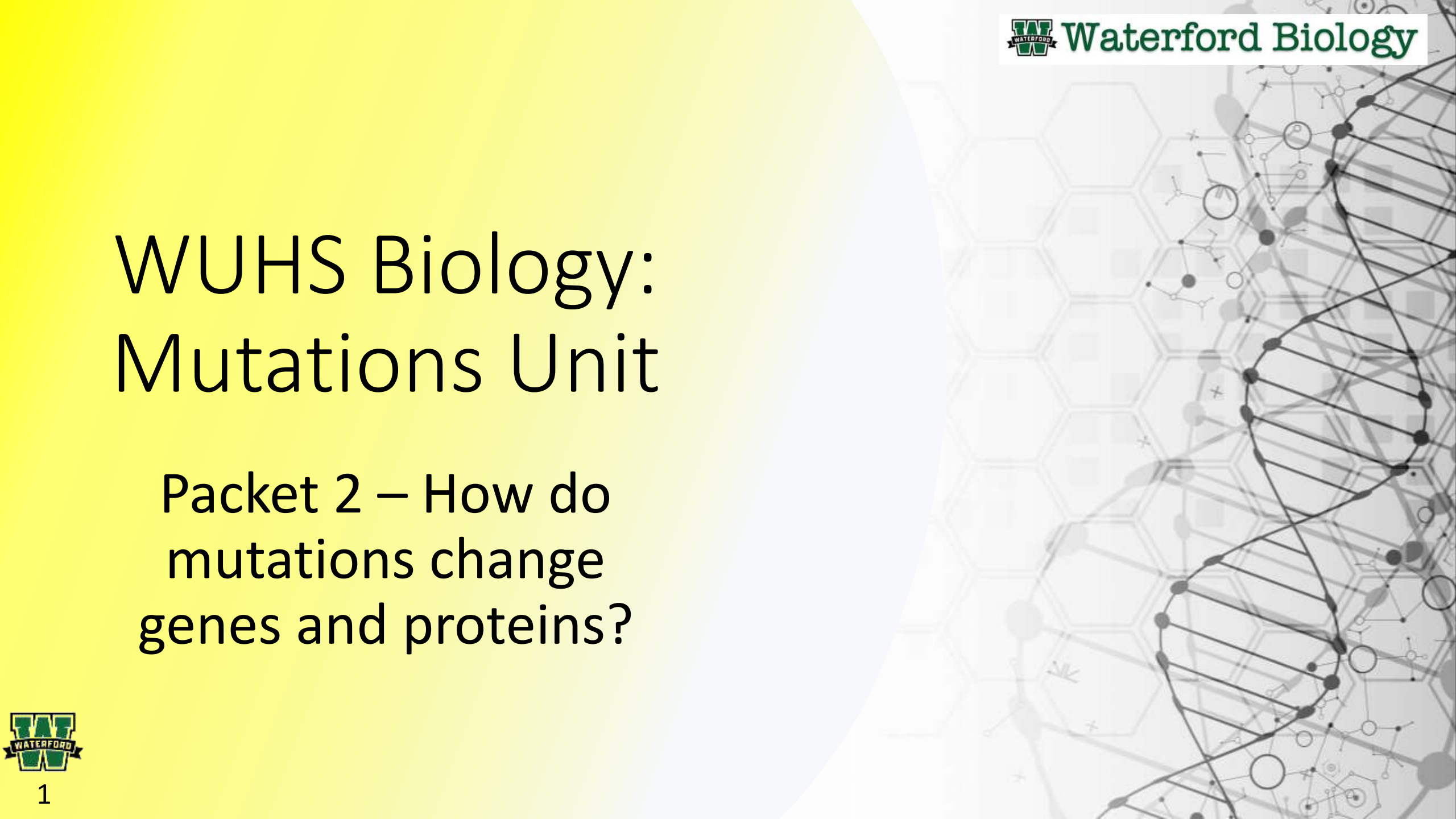
Waterford Biology

WUHS Biology: Mutations Unit

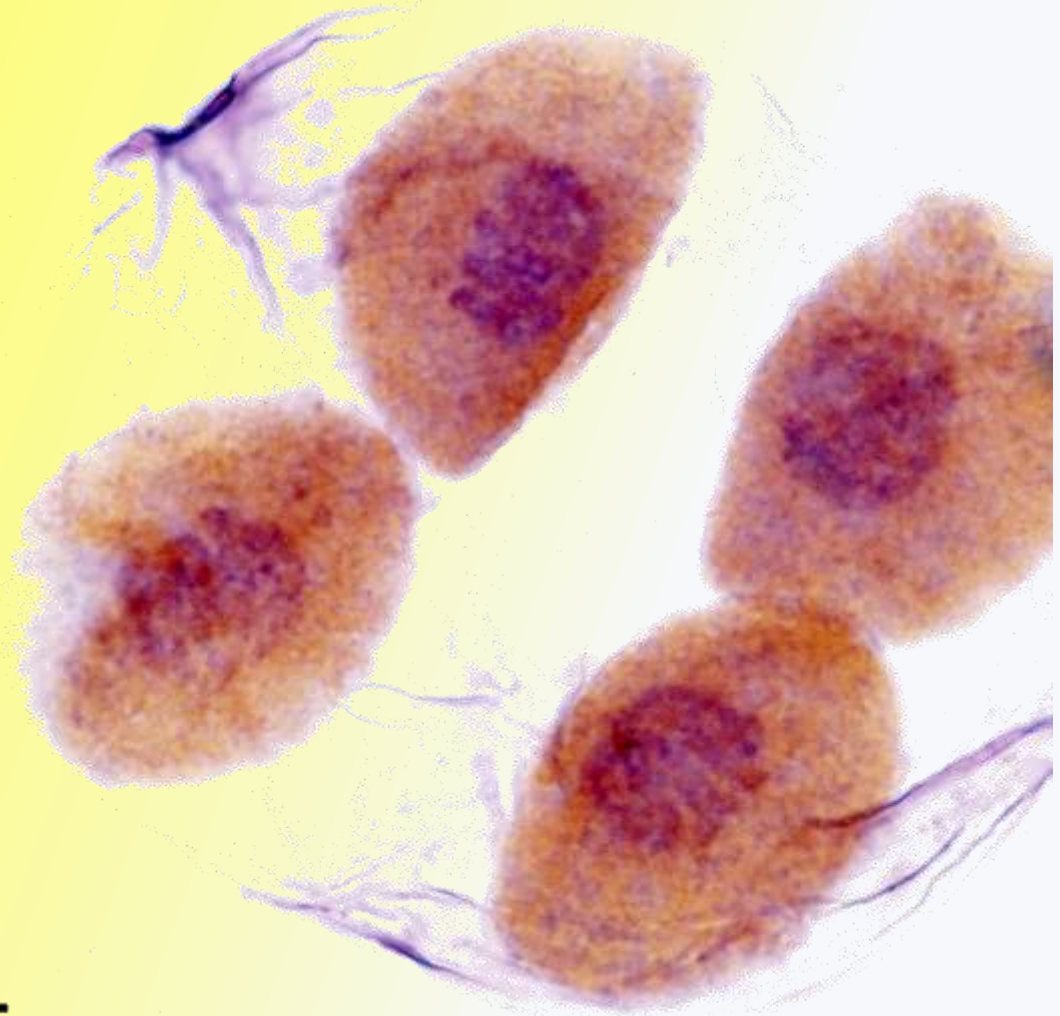
Packet 2 – How do mutations change genes and proteins?



Mutations Unit – Packet 2 Driving Question

- **Driving Question: How do mutations change genes & proteins?**
- What kinds of mutations can occur?
- Why do mutations occur?
- What are the impacts of different mutations?

Image Source: : Pixabay



Recap of the Previous Units

What have we learned so far?

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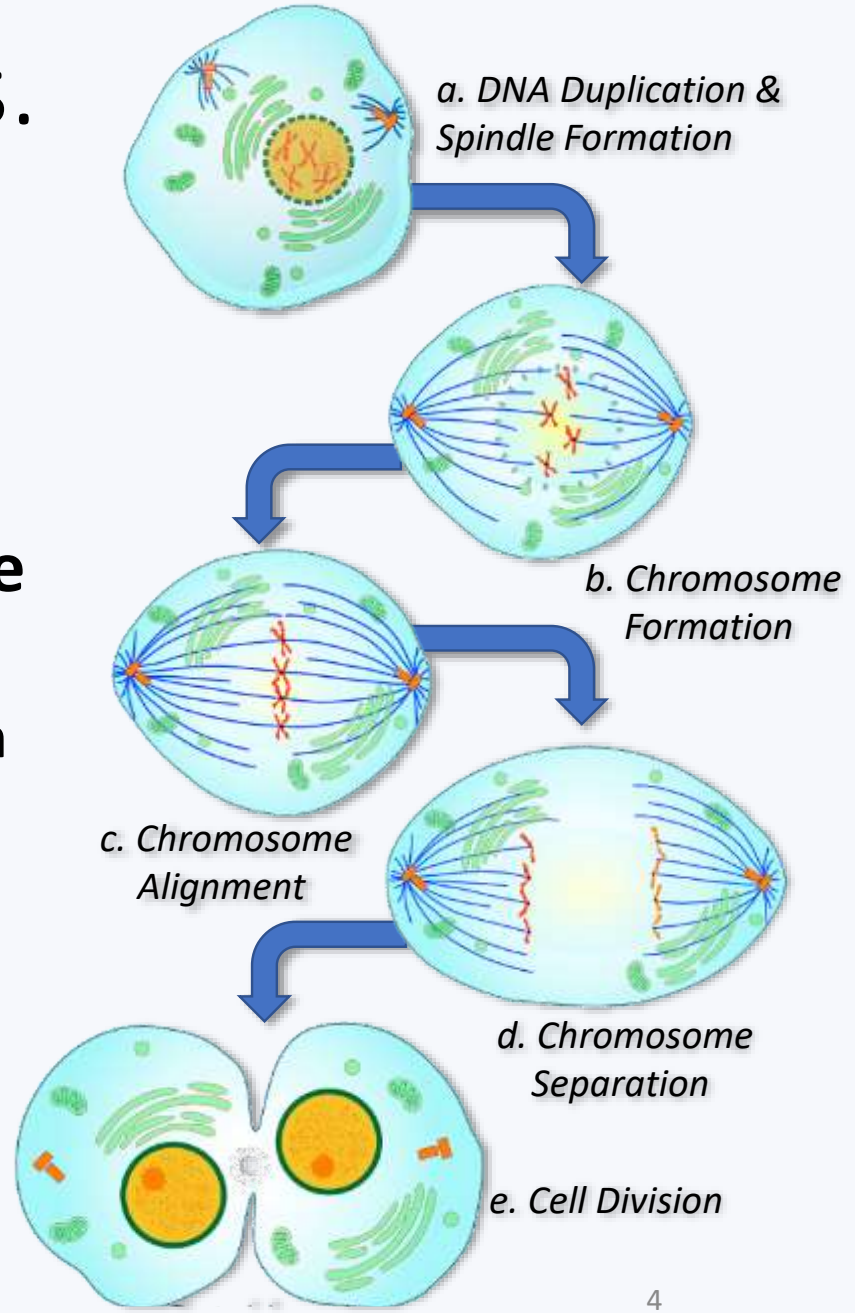
Recap: How DNA is used by cells.

- **An organism's genes (segments of DNA) determine how their cells assemble proteins.**
  - Proteins are primarily responsible for the observable traits of an organism.
- **When a cell undergoes mitosis (cell division), the DNA must be duplicated.**
  - The double-stranded DNA is opened by helicase; each strand of DNA is then duplicated by polymerase.
- **This DNA is then packaged into chromosomes and evenly divided between the two cells.**
  - Each cell must have its own copy of DNA to assemble its own proteins.

Image Source: Wikimedia



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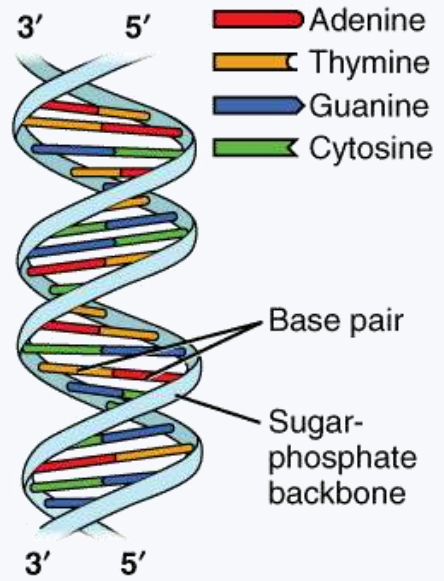
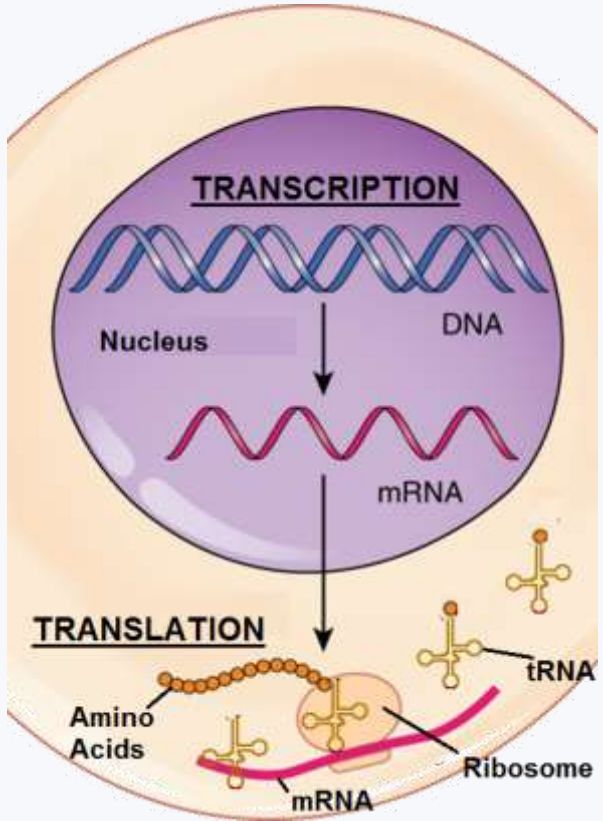
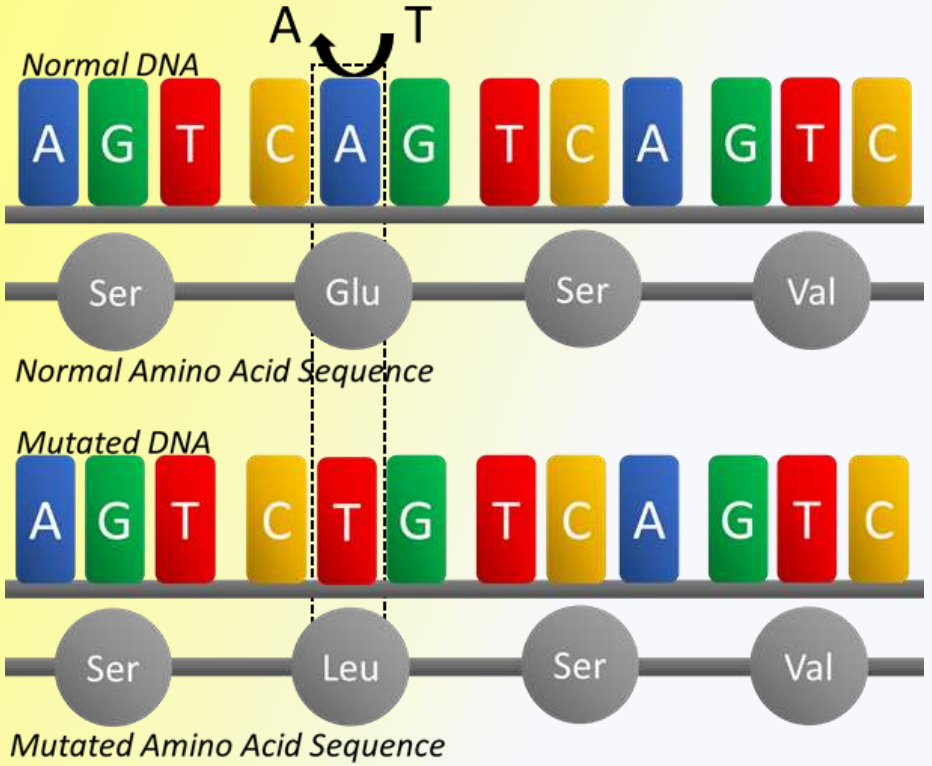
DNA & Proteins Unit Recap

- **DNA is a macromolecule made from repeating chains of nucleotides.**
  - Nucleotides consist of phosphate, sugar, and base molecules.
  - DNA provides instructions for assembling proteins.
- **DNA contains four bases: G, C, A, and T.**
  - G always binds to C. A always binds to T.
  - Groups of 3 bases (codons) code for specific amino acids.
  - The order of codons determines the order of amino acids.
- **During transcription, a copy of DNA (mRNA) is made.**
- **During translation, a ribosome assembles a protein based on the information copied in mRNA.**
  - The properties of amino acids determine the shape & function of the protein.







Types of Mutations

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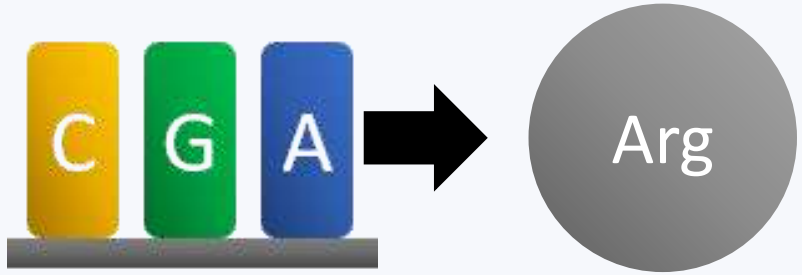
Mutations = Mistakes

- **Sometimes mistakes happen when DNA is being replicated or copied.**
  - A mutation occurs when DNA undergoes a permanent but unintentional change through the loss, addition, or switching of at least one base.
  - For example, if a CAG codon is mistakenly copied as CGA, it will code for a different amino acid.

- **Changing a codon in a gene can change the order in which amino acids are assembled.**
  - This can change the shape of that protein, which could change the function of that protein.



- **There are several different kinds of mutations.**
  - Mutations can range from only affecting a single base to changing multiple genes on a chromosome.
  - The more codons affected by a mutation, the more a mutation changes the shape and function of a protein.

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Acquired Mutations

- **Mutations can be either acquired or hereditary.**
- **<u>Acquired mutations</u> occur sometime during the life of an organism (*i.e., these mutations did not initially exist in an organism's DNA in their early cells*).**
  - Acquired mutations are only found in some cells in an organism.
  - Acquired mutations are usually not passed on to offspring.
- **Acquired mutations can be caused by environmental factors or by replication mistakes during mitosis.**
  - Environmental factors that cause mutations are called <u>mutagens</u>.
    - *Potential sources of mutagens include UV radiation (sunlight), processed food, cigarette smoke, and X-rays.*
  - Acquired mutations can also result from mistakes that occur as DNA is duplicated by polymerase during mitosis.

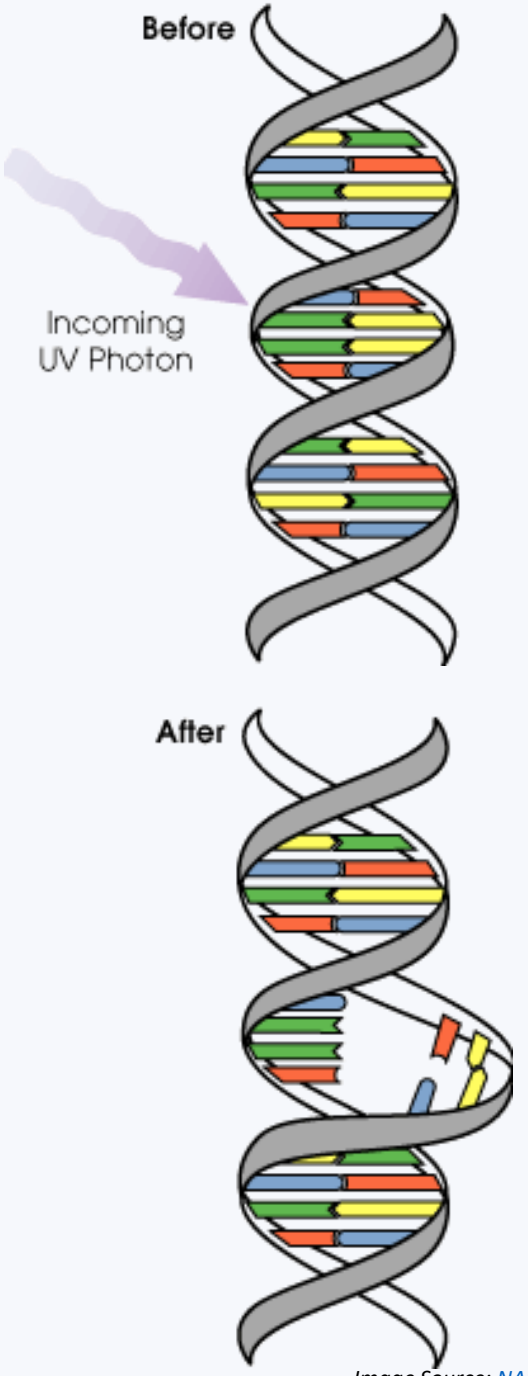
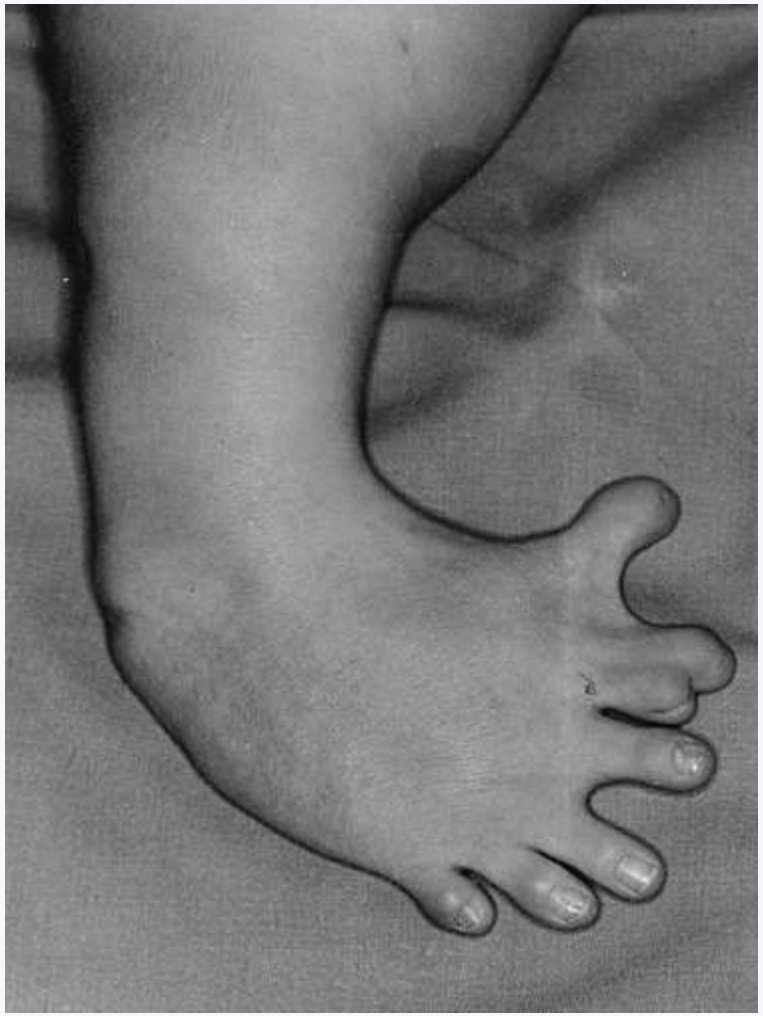


Image Source: NASA

Case Study: Thalidomide

- **Thalidomide was a drug used to treat nausea in pregnant women in the mid-1900s.**
  - Researchers soon discovered that thalidomide treatment caused severe birth defects in thousands of children.
- **Thalidomide caused *acquired* mutations – the DNA of affected children changed as they were developing in the uterus because of the drug.**
  - The mutations that affected these children were not found in the sperm or egg cells of their parents.
  - Instead, the mutations developed after fertilization.

Source: Wikimedia

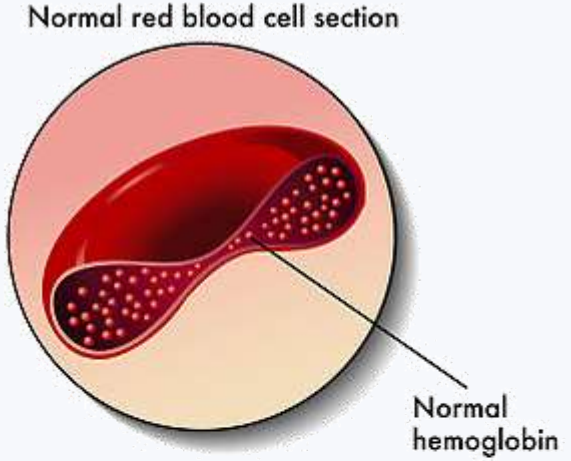


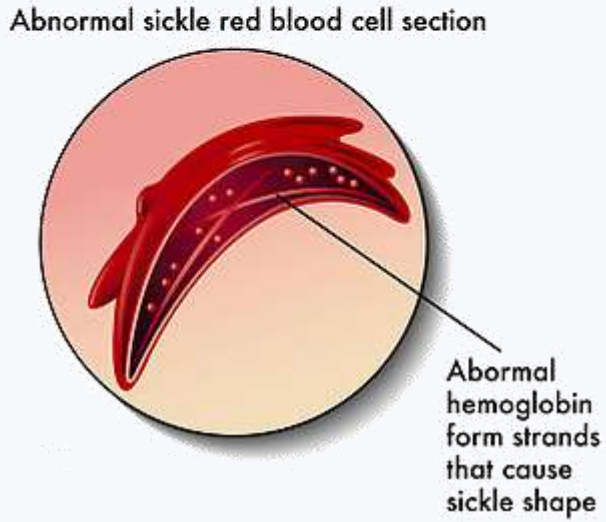
Thousands of children had incidences of birth defects as a result of thalidomide.

Source: Wikimedia

Hereditary Mutations

- **<u>Hereditary mutations</u> are those that can be passed on to subsequent generations and are present throughout the life of an organism.**
  - These mutations are found in *every* cell of an organism.
- **A mutation becomes hereditary if found in sperm or eggs cells.**
  - During fertilization, a sperm and egg cell combine their DNA.
  - If either the egg or the sperm cell carry a mutation, all cells that divide from this cell will also replicate this mutation.
- **Hereditary mutations can result from different causes.**
  - Some hereditary mutations result from copying errors in meiosis.
  - Some hereditary mutations occur during crossing over in meiosis.
    - *For example, bases of a gene can be lost or added when chromosomes exchange segments during crossing over.*
  - Mutagens can also change DNA in sperm and egg cells, resulting in hereditary mutations.





Genetic diseases, such as sickle anemia, are usually examples of hereditary mutations because they are usually inherited from parents.

Case Study: Double Muscle Cattle

- **“Double muscling” is a genetic condition that can occur in cattle.**
  - Animals affected by this condition inherit a mutation in their gene for the myostatin protein.
  - This protein regulates muscle development and limits the rate at which animals can develop muscle fibers.

- **Cattle with the mutated myostatin protein are born with 2-3x as many muscle fibers.**
  - This results in visibly greater muscle growth compared to unaffected cattle.
  - This is a hereditary mutation - it is passed on from parents and affects all cells of the organism.

Image Source: Pixabay

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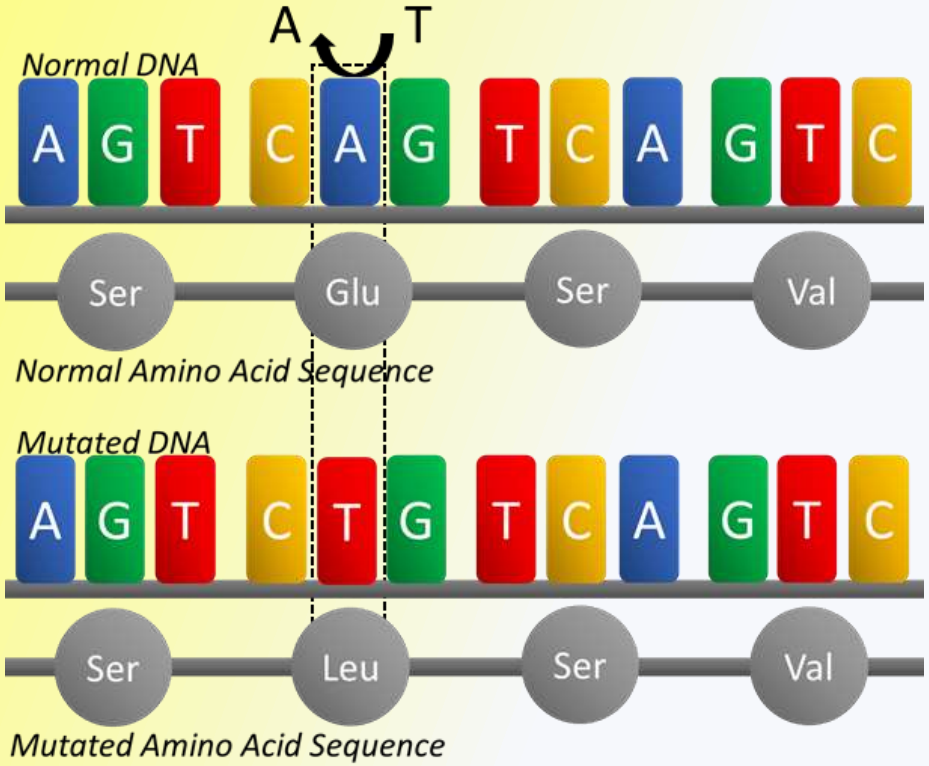

Mutations Mechanisms – How DNA is Altered

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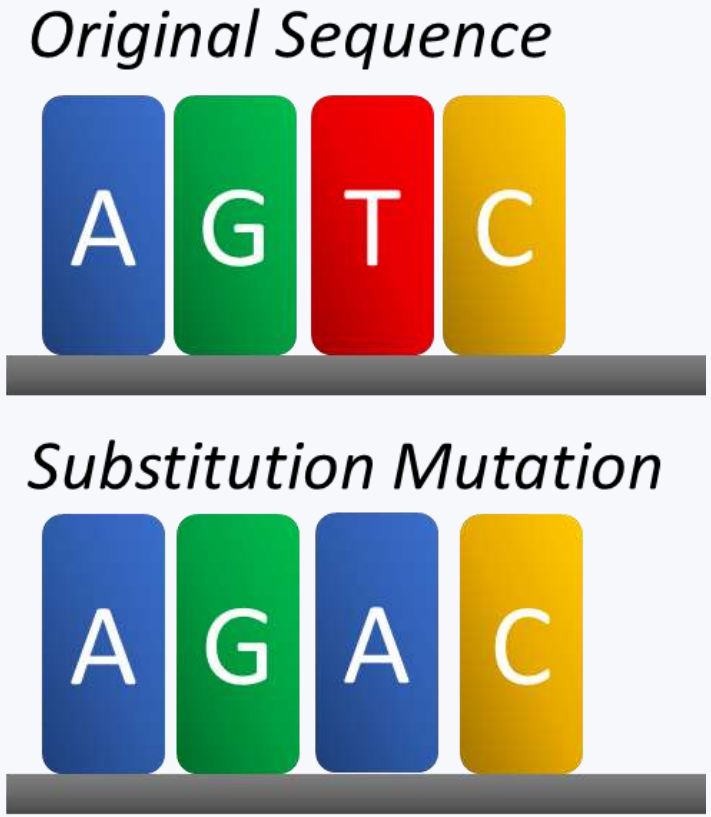
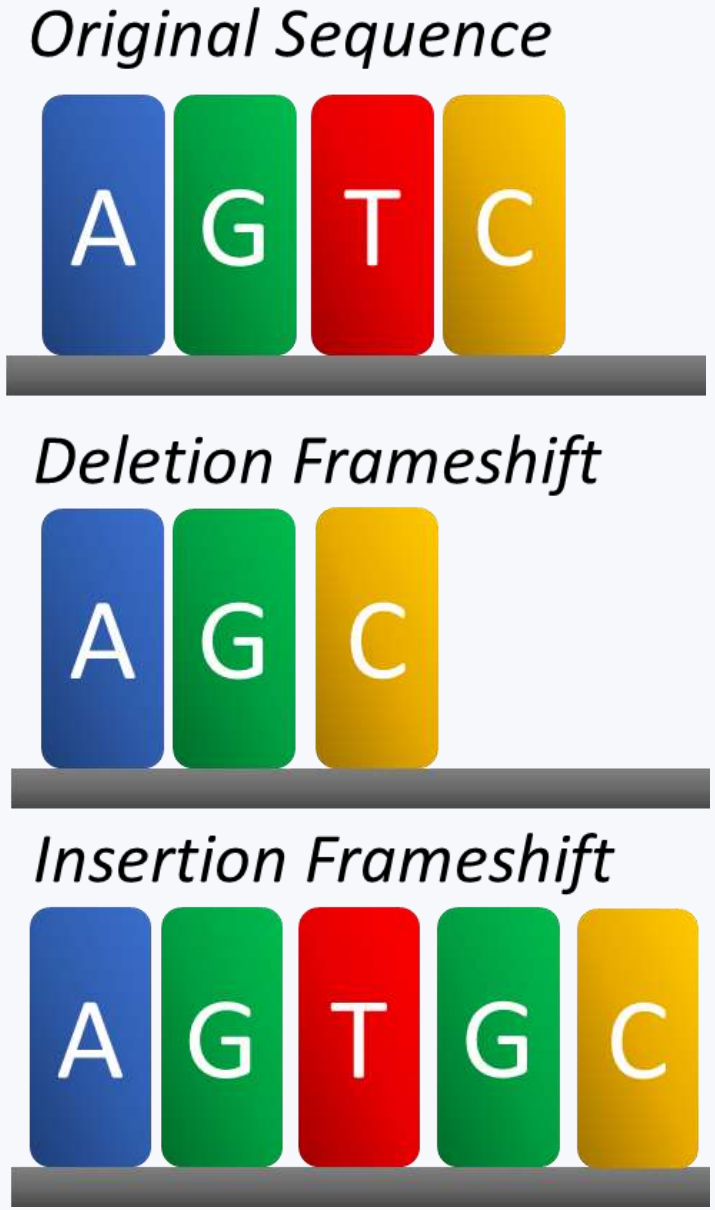
Mutations Mechanisms

- **A <u>substitution mutation</u> (*or point* mutation) occurs if one base is replaced by another base.**
  - In these mutations, usually only one codon is changed.
- **Some substitutions have no impact.**
  - For example, changing AAG to AAA still codes for lysine).
- **If a substitution results in a different amino acid, this can change the protein shape and function.**
  - If a substitution creates a stop codon, protein assembly will stop prematurely, resulting in only a partial protein.

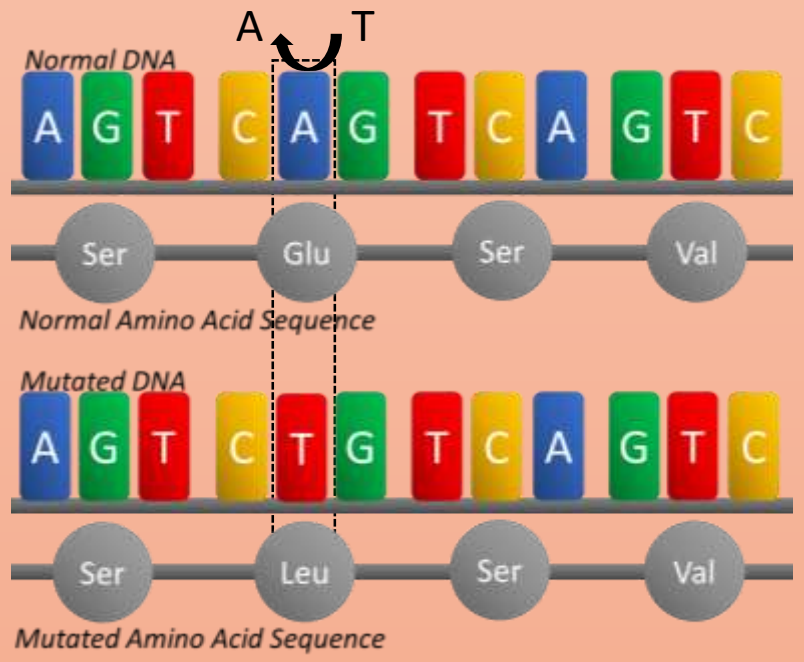


Mutations Mechanisms

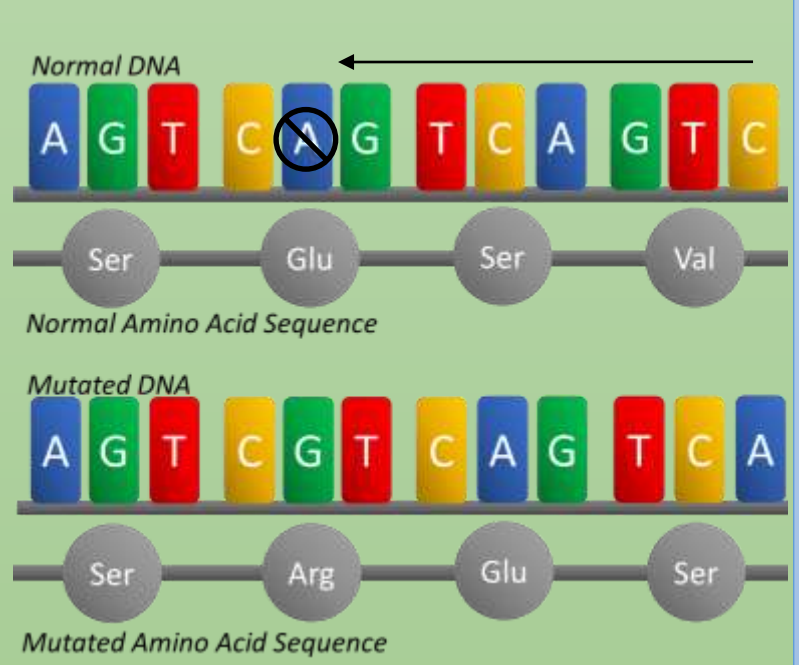
- **A <u>frameshift mutation</u> changes every codon that occurs after the mutation due to the insertion or deletion of a base.**
  - Frameshift mutations tend to have the biggest impact on protein assembly because they affect codons and amino acids after the mutation.
- **Frameshift mutations can be caused by insertion or deletions.**
  - <u>Insertions</u>: bases are added to genes.
  - <u>Deletions</u>: bases are removed from genes.



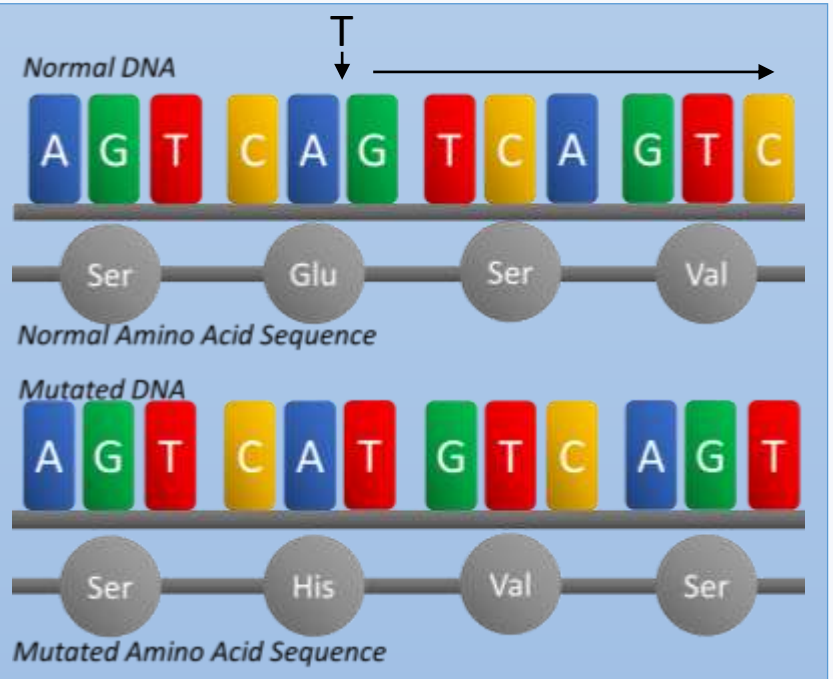
Mutations Mechanisms w/ Effects on Translation



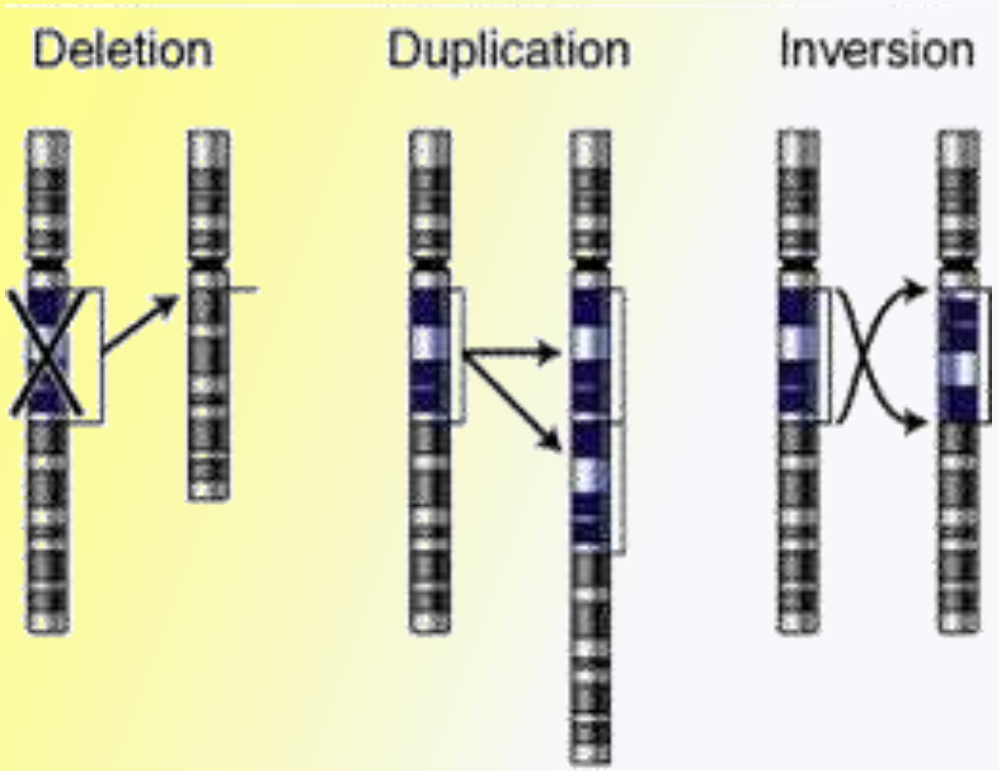
- **<u>Substitution</u>: one base is swapped for another.**
- **All other bases are unaffected.**
- **Other codons are unaffected (*unless a stop codon is created*).**



- **<u>Deletion Frameshift</u>*:* one base is removed from the gene.**
- **All other bases shift one space backward.**
- **All subsequent codons change.**



- **<u>Insertion Frameshift</u>: one base is added to the gene.**
- **All other bases shift one space forward.**
- **All subsequent codons change.**



Chromosomal Mutations – Changing Multiple Genes

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Chromosomal Mutations

- **Sometimes mutations can involve multiple genes.**
  - These are known as chromosomal mutations because they involve changes in the structure of a chromosome.

- **Chromosomes can be mutated in four key ways.**
  - Deletion: when part or all of the genes in a chromosome are lost.
  - Duplication: when all or part of a chromosome is unintentionally replicated.
  - Inversion: when the order of genes on a chromosome is reversed.
  - Insertion/Translocation: when part of one chromosome is unintentionally inserted onto another chromosome.

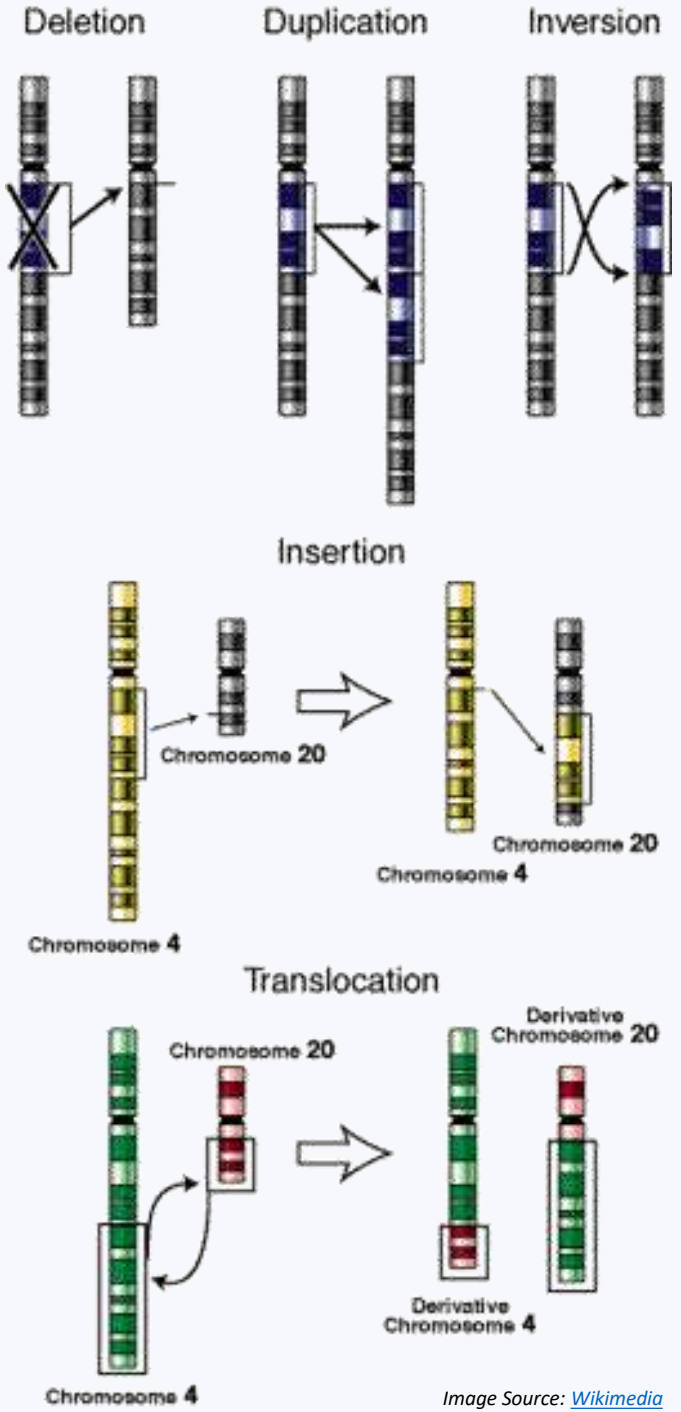

Image Source: Wikimedia

Effects of Chromosomal Mutations

- **The effects of chromosomal mutations can vary.**
  - Some chromosomal mutations may have no effect on a person's health; some chromosomal mutations can result in significant health problems.
  - The impact of these mutations depend on how many genes are affected, which genes are affected, whether gene function is interrupted, and whether any genes are gained or lost.

- **One common example of a chromosomal mutation is Down's syndrome.**
  - Down's syndrome results from errors during meiosis, resulting in gametes (sperm or egg cells) with three copies of the 21st chromosome.
  - As a result, individuals with Down's syndrome possess 47 chromosomes instead of 46.

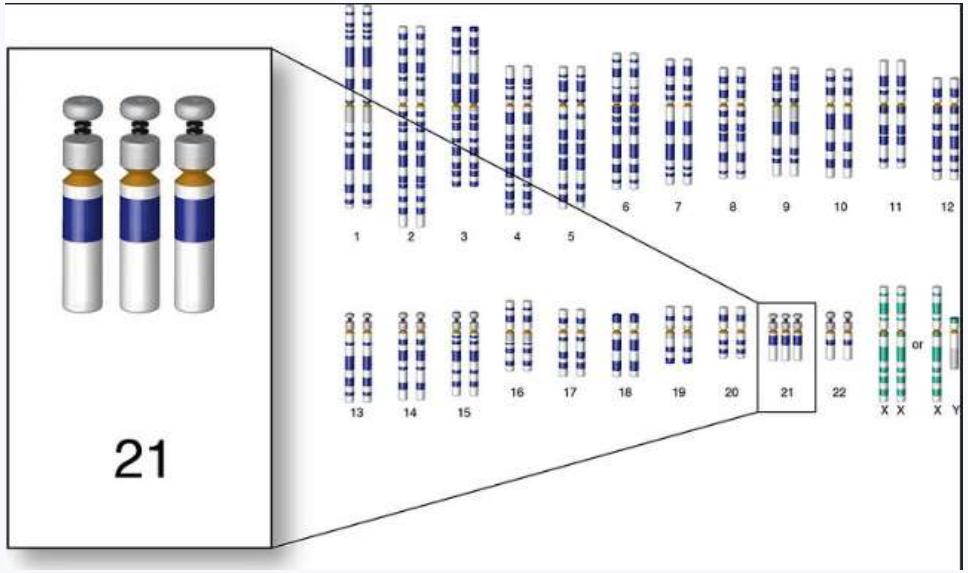

Image Source: Flickr

Revising Our Claims

- **Revisit your ideas from Part 1.**
  - How could you improve your responses to our Driving Questions?
- **Driving Question: How do mutations change genes & proteins?**
- What kinds of mutations can occur?
- Why do mutations occur?
- What are the impacts of different mutations?

Image Source: : Pixabay

Looking Ahead: Part 3 Investigation

- **In Part 3 you will be using a computer simulation to explore different kinds of mutations and their impacts of protein assembly.**

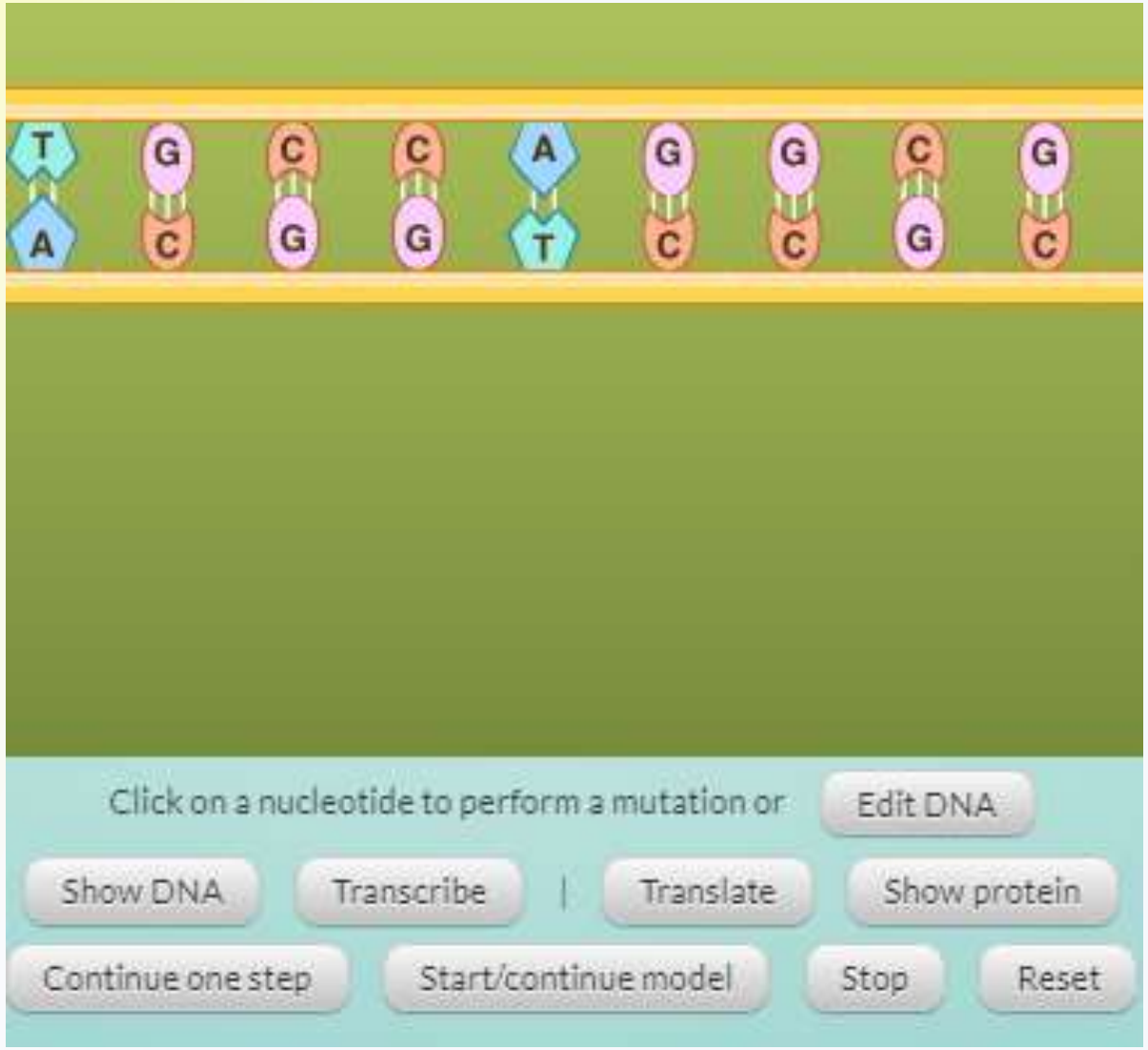

Image Source: Biology Corner

Key Points

- **A mutation occurs when DNA undergoes a permanent but unintentional change through the loss, addition, or switching of at least one base.**
  - This changes codons, which can change the order of amino acids, which can change the shape and function of a protein.
- **Acquired mutations occur sometime during the life of an organism.**
  - Acquired mutations are only found in some cells and are not passed on to offspring.
  - These mutations can be caused by replication errors during mitosis or by mutagens (environmental factors like UV radiation that can cause mutations).
- **Hereditary mutations are those that can be passed on to subsequent generations and are present throughout the life of an organism.**
  - These mutations are found in every cell of an organism and can be passed on to offspring.
  - These mutations can be caused by replication errors during meiosis, by crossing over during meiosis, or by mutagens that affect the DNA of sperm or egg cells.

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Key Points

- **A substitution mutation (or point mutation) occurs if one base is replaced by another base.**
  - Only one codon is changed, which can change the amino acid it codes for.
  - Unless the substitution mutation results in a stop codon, these mutations generally do not affect the other codons or amino acids.
- **A frameshift mutation changes every codon that occurs after the mutation due to the insertion or deletion of a base.**
  - Frameshift mutations tend to have the biggest impact on protein assembly because they cause all the other amino acids to change after the mutation.
  - Frameshift mutations result from the insertion or deletion of a base.
- **Chromosomal mutations involve changes in the structure of a chromosome and affect multiple genes.**
  - Chromosomal mutations can be caused by deletion or duplication of genes.
  - They can also result from inversions (reversal of gene order) or insertions/translocations (movement of genes to new chromosomes).

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Key Vocab

- **Mutation - when DNA undergoes a permanent but unintentional change through the loss, addition, or switching of at least one base.**
- **Acquired mutations occur sometime during the life of an organism and cannot be passed on to offspring.**
- **Mutagens - environmental factors that cause mutations.**
- **Hereditary mutations are those that can be passed on to subsequent generations and are present throughout the life of an organism.**
- **A substitution mutation occurs if one base is replaced by another base.**
- **A frameshift mutation changes every codon that occurs after the mutation due to the insertion or deletion of a base.**
- **Chromosomal mutations affect multiple genes and involve changes in the structure of a chromosome.**

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