

GENETICS

LESSON 9: MUTATIONS

Learning Outcomes

- I can define what a mutation is.
- I can explain when a mutation is beneficial, neutral or harmful.

Species

- a group of individuals that can interbreed and produce fertile offspring
- share a common DNA base that has slight variations in some of the DNA sequences (alleles)

Mutations

- a permanent change in the DNA sequence of an individual
- may occur in:
 - somatic cells – normal body cells that may result in a change in that individual
 - germ line cells – gamete (sperm & egg) producing cells that may result in a change in all of an individual's offspring
- only germ line mutations can result in a change to a species as they are the source of new alleles
- mutations can be:
 - beneficial – aids in survival (ex: better camouflage)



- harmful – results in death or decreased survival ability (ex: third hind leg on frogs)



- neutral – changes in the DNA sequence does not change the organisms (ex: in non-reading portion of DNA) or changes that do not influence survival rates (ex: dichromatic eyes)



Effects of Mutations

- remember that genes in DNA code for the proteins that carry out many of the functions in your body – structural proteins, hormones, antibodies, etc.
- when a mutation changes the sequence of a gene (DNA sequence), it can result in:
 1. Point Mutation (or Substitution)

- occurs when one base is changed for a different base
- can result in:
 - silent mutation – order of amino acids after translation does not change

	Original Strand	Mutated Strand
DNA	TAC GCG AAT AGT ACT	TAC GCG AAT AGG ACT
mRNA	AUG CGC UUA UCA UGA	AUG CGC UUA UCC UGA
Amino Acid	met – arg – leu – ser – stop	met – arg – leu – ser – val

- missense mutation – single nucleotide change results in different amino acid

	Original Strand	Mutated Strand
DNA	TAC GCG AAT AGT ACT	TAC GCG AAT GGT ACT
mRNA	AUG CGC UUA UCA UGA	AUG CGC UUA CCA UGA
Amino Acid	met – arg – leu – ser – stop	met – arg – leu – pro – stop

- nonsense mutation – mRNA codes for an early stop codon

	Original Strand	Mutated Strand
DNA	TAC GCG AAT AGT ACT	TAC GCG ATT AGT ACT
mRNA	AUG CGC UUA UCA UGA	AUG CGC UAA UAA UGA
Amino Acid	met – arg – leu – ser – stop	met – arg – stop

2. Frameshift Mutation

- a change in the reading frame of RNA caused by adding or removing bases
- results in:
 - Deletion Mutation – loss of a base or bases causing a shortening of the amino acids sequence

	Original Strand	Mutated Strand
DNA	TAC GT G CGA AAT GAT	TAC GGC GAA ATG AT
mRNA	AUG CAC GCU UUA CUA	AUG CCG CUU UAC UA
Amino Acid	met – his – ala – leu – leu	met – pro – leu – tyr

- Insertion Mutation – addition of a base or bases causing a lengthening of the amino acid sequence

	Original Strand	Mutated Strand
DNA	TAC GTG CGA AAT GAT	TAC GTA GCG AAA TGA T
mRNA	AUG CAC GCU UUA CUA	AUG CAU CGC UUU ACU A
Amino Acid	met – his – ala – leu – leu	met – his – arg – phe – thr

- frameshift mutations usually result in more harmful effects since there is often a greater change in the amino acid sequence

Causes of Mutation

- can occur spontaneously during cell division
- may be caused by external influences → mutagens are substances or events that can cause a change in DNA
- two main categories of mutagens
 1. Physical Mutagens
 - cause a physical change in the sequence of DNA in an organism
 - examples: UV light, X-ray radiation
 2. Chemical Mutagens
 - caused by chemicals that enter a cell and chemically react with DNA
 - examples: nitrites, gasoline fumes
- Carcinogens
 - physical or chemical mutagens that result in the formation of cancer

<https://www.youtube.com/watch?v=1v55yg0RfoY>

Sickle cell anemia is an example of a missense mutation

https://www.ted.com/talks/amber_m_yates_how_this_disease_changes_the_shape_of_your_cells/transcript?language=en#t-240861