

MUTATIONS



Cells Make Mistakes!

- Mutations – changes in the genetic material

3 Types of Mutations: *Point*

1. Substitutions – 1 nucleotide is exchanged for another
 - Only affect 1 nucleotide
 - E.g. TAC GCA TGG
TAC GTA TGG
 - Results in a change in 1 amino acid

3 Types of Mutations: *Frameshift*

2. Additions – adding nucleotides into a sequence

- E.g. TAC GCA TGG
TAC G**C**CA TGG

3. Deletions – deleting nucleotides from a sequence

- E.g. TAC G**C**A TGG
TAC GA TGG

****Additions and Deletions have more serious consequences because it shifts the sequence and changes more than 1 codon and therefore amino acid****

What causes mutations?

- ⦿ DNA does not copy correctly
- ⦿ Environmental factors:
 - Chemicals
 - Radiation
 - Sunlight (temperature)
 - Smoking

Effect of Mutations

- ⦿ Most have little or no effect on gene expression
- ⦿ Ones that do have an effect- it is usually dramatic
 - Could cause genetic disorders
 - Could benefit the organism

*Mutations in the zygote have **SERIOUS** consequences*

Gene Regulation

- Only a fraction of genes in a cell are expressed at a given time
- Expressed gene – one that is transcribed into RNA

How does a cell determine which genes will be expressed and which will be silent?

- DNA-binding proteins attach specific DNA sequences
 - Help regulate gene expression

Throwback:

- Stem Cells – unspecialized ‘generic’ cells
- Become specialized (e.g. muscle or nerve) by the process of transcription
 - i.e. transcription of different genes leads to the synthesis of different proteins

Remember Cell Differentiation leads to Cell Specialization!

- ⦿ Different types of cells have specialized functions
- ⦿ Cells must communicate in order to coordinate the activity of each cell