GENETIC MUTATIONS

EQ: How do changes to our DNA affect us?

What are genetic mutations?

- All genetic disorders are caused by a mutation
- Mutation: A change in the genetic base-code for a protein.
- A mutation happen regularly and can occur at almost any stage in development
  - DNA replication ,mitosis, meiosis, chromosome separation.
- Environmental factors can lead to mutations as well.
- Many mutations are repaired by enzymes

What are the types of mutations?

Beneficial Mutations: Mutations that help your chances of survival

Dark skin: + resistant to sunburn

- generate less Vitamin D

*Increased survival in sunny environments, like the equator.

Light skin: + generate more Vitamin D

- prone to sunburn

*Increased survival in less sunny environments.

Harmful: Mutations that decrease chances of survival

Ex: genes coding for proteins that control growth; this results in un-controlled cell growth.

Neither: This does not increase or decrease a person's chance of survival.

Ex: Attached earlobes

What happens if your body doesn't work exactly as it is supposed to?

- **Genetic Disorders result when there is a change in your genes that changes the way your body functions**.
- Sometimes the change can be so large that your body cannot function.

Common Genetic Disorders

Disorder	Mutation
• Sickle-Cell Anemia	• Change in one base pair
• Down Syndrome	• Chromosomes do not separate evenly in meiosis
• Lactose Intolerance	• Gene does not produce particular protein that digests sugars in milk
• Colorblindness	• Multiple genes that allow us to see color are not coded for (on X chromosome)

Common Genetic Disorders

Disorder	Mutation
• Muscular Dystrophy	• Two recessive genes (passed from parents or develops over time)
• Alzheimer Disease	• Multiple genes and environmental effects; gene coding for protein that interferes with nerve shape is over produced
• Cancer	• Multiple genes and environmental effects; changes in genes that code for growth

What is cancer?

- Like Alzheimer disease, **cancer is caused by both genetic mutations and environmental effects.**
  - Environmental effects, such as excess sun exposure, are thought to be a partial cause of genetic mutations.
- **There are many types of cancer, but they all involve un-controlled cell growth.**

Lance Armstrong,
7 time winner of the Tour de France,
Testicular cancer

Nancy Regan,
Former First Lady,
Breast cancer

Bob Marley,
reggae musician,
Died of skin cancer in 1981

What is cancer?

- **Mutations occur in three types of genes to cause cancer** (un-controlled cell growth):
  - **1. Genes that promote normal cell growth are mutated and function at a higher level.**
  - **2. Genes that stop cell growth are mutated and do not function.**
  - **3. Genes which code for proteins that repair DNA mutations are mutated.**
    - Genes that code for proteins that check for mutations in DNA act as a second protection against mutation.
    - Without this check system, cancer is extremely deadly.