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

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sickle-Cell Anemia</td>
<td>Change in one base pair</td>
</tr>
<tr>
<td>Down Syndrome</td>
<td>Chromosomes do not separate evenly in meiosis</td>
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<tr>
<td>Lactose Intolerance</td>
<td>Gene does not produce particular protein that digests sugars in milk</td>
</tr>
<tr>
<td>Colorblindness</td>
<td>Multiple genes that allow us to see color are not coded for (on X chromosome)</td>
</tr>
</tbody>
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Common Genetic Disorders

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<tr>
<th>Disorder</th>
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<tr>
<td>Muscular Dystrophy</td>
<td>Two recessive genes (passed from parents or develops over time)</td>
</tr>
<tr>
<td>Alzheimer Disease</td>
<td>Multiple genes and environmental effects; gene coding for protein that interferes with nerve shape is over produced</td>
</tr>
<tr>
<td>Cancer</td>
<td>Multiple genes and environmental effects; changes in genes that code for growth</td>
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</tbody>
</table>
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.