Assignment Number:\_\_\_\_\_\_\_\_\_

Name:\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Hour:\_\_\_\_\_\_\_\_\_\_\_

**Mutations: The Basis of Genetic Change**

**Mutation** – a change in the \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ or \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ of genetic material of an organism, can be caused by \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ or \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ incorrectly

* Mutations can occur \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ during the cell cycle when DNA is not copied correctly, be \_\_\_\_\_\_\_\_\_\_\_\_\_\_ from a \_\_\_\_\_\_\_\_\_\_ or developed due to the \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (such as those caused by radiation or chemicals).
* Mutations can be expressed differently and affect differing amounts of DNA based on what the mutation actually is. A mutation can only be as \_\_\_\_\_\_\_\_\_\_ as one piece of DNA long or an entire \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (or more!).
* Mutations can have many possible effects. They could be small and have \_\_\_\_\_ effect, they could \_\_\_\_\_\_\_\_\_\_\_ your cells, or they could \_\_\_\_\_\_\_\_\_\_\_ your cells.
* Everyone carries many \_\_\_\_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_\_\_ that are not always \_\_\_\_\_\_\_\_\_\_\_\_\_ or harmful
  + Inbreeding can cause them to get expressed
* Mutations that occur in \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (sex cells) are the \_\_\_\_\_\_\_\_\_ ones that get passed on to the \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_.

Different Types of Mutations

1. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ – when \_\_\_\_\_\_\_\_\_\_\_\_\_ DNA is added to the sequence
2. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ – when DNA is deleted (\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_) from the sequence
3. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ – one segment of DNA \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ another

All of these types of mutations will be passed on to every cell that develops off of the mutated cell. BUT remember \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ that occur in \_\_\_\_\_\_\_-sex cells \_\_\_\_\_\_\_\_\_\_\_\_\_ be passed on to the next generation (offspring).

**How can we identify mutations?**

We can analyze small pieces of DNA but replicating them and running them through gel electrophoresis. We can analyze whole chromosomes by looking at \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_.

**Karyotype** – is an organized \_\_\_\_\_\_\_\_\_\_\_\_\_\_ of a person’s \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_. Chromosomes are arranged and numbered by \_\_\_\_\_\_\_\_\_\_\_ (from largest to smallest). This genetic picture makes it easy to recognized chromosomal mutations.

*Let’s review quick: A normal human has how many chromosomes total?*

*How many DIFFERENT chromosome pairs does a normal human have?*

***Homologous Pair of Chromosomes*** *– have the same \_\_\_\_\_\_\_ in the same \_\_\_\_\_\_\_\_\_\_\_\_*

*Each organism gets one chromosome from their \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_, one from their \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ to make up the pair.*

**Chromosomal Mutation Disorders:**

1. **Down Syndrome** – also know as, trisomy 21, an individual with Down Syndrome has \_\_\_\_\_\_\_ chromosomes because instead of having only \_\_\_\_\_\_\_\_ 21st chromosomes they have \_\_\_\_\_\_\_\_\_\_
   1. Children who have down syndrome often have mental disabilities and distinct facial features
2. **Turner’s Syndrome** – an individual with Turner’s syndrome only has \_\_\_\_ chromosomes, because they are lacking an entire \_\_\_\_\_\_\_ chromosome, these girls only have \_\_\_\_\_\_ X and experience abnormalities such as non-working ovaries, webbed necks, short stature, broad chests, low ears and often many additional internal abnormalities
3. **Klinefelter’s Syndrome** – an individual who has \_\_\_\_\_\_\_\_ chromosomes because they are a male with two \_\_\_\_ chromosomes instead of one (\_\_\_\_\_\_), those affected with the syndrome tend to have small sexual organs and reduced fertility

**Gene Mutation Disorders:**

1. **Sickle Cell Anemia (\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_)** - caused by \_\_\_\_\_\_\_\_ coding for a \_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_ (hemoglobin), cells are \_\_\_\_\_\_\_\_ shape, stiffer and sticky. They tend to \_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_ in the blood vessels of limbs and organs.
2. **Hemophilia** – caused by genes coding for a defective \_\_\_\_\_\_\_\_\_\_ (autoantibody), \_\_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ that slows the blood \_\_\_\_\_\_\_\_\_\_\_\_\_ process
3. **Cystic Fibrosis** – caused by genes coding for a defective \_\_\_\_\_\_\_\_\_\_\_\_\_ (channel protein that lets chloride out of cells), causes a \_\_\_\_\_\_\_\_\_\_\_\_\_ of thick, sticky \_\_\_\_\_\_\_\_\_\_\_ that can damage \_\_\_\_\_\_\_\_\_\_\_\_\_