Workshop notes 4 : **Mutations**

**Mutation** = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_  

A mutation can occur in an individual \_\_\_\_\_\_\_\_\_\_\_\_\_\_

 - results in a single changed \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

- cystic fibrosis a mutation in the protein that makes a type of ion channels in cell membrane

- bacterial \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ to antibiotics is an example of a beneficial gene mutation

A mutation can occur in a \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

 - a chromosome contains many \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

 - chromosomal mutations affect many proteins

If a mutation occurs in a **\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_**, the mutation will be passed to offspring

 Examples: Down Syndrome, Edward’s Syndrome, Cri-du-Chat

**Causes of Mutations**:

1. Environment: Can be caused by \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_- a physical or chemical cause of mutation. Examples: UV light, radiation, drugs, and benzene.
2. Mutagens are often also \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ – anything that causes cancer
3. Can be \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_, random events.

 - mutations occur in 1/100,000 DNA replications (DNA \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_)

* Mutations do not have to be bad (evolution)

**Gene Mutations**:

1. Point Mutations

* A single \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ is altered. Can change one amino acid in a \_\_\_\_\_\_\_\_\_
* Mil**k** – Mil**e**
* GGAC**A**ATCA GGAC**C**ATCA

 proline -valine-serine proline-glycine-serine

**\*\*\*ONLY ONE AMINO ACID CAN BE AFFECTED AS A RESULT OF A POINT MUTATION!\*\*\***

2. Frameshift Mutations

* A \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ is either inserted or deleted from a \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_.

all of the triplets from the point of \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ onward will be changed

* An \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ occurs when a nucleotide is added to a gene

 Example: A nucleotide is inserted

The fat cat ate the rat

 The f**a**a tca tat eth era t

 -the extra nucleotide *\_\_\_\_\_\_\_\_\_\_\_\_\_* all of the triplets that follow

* A \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ occurs when a nucleotide is removed from a gene.

 Example: A nucleotide is \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

 The fat cat ate the rat

 Thf atc ata tet her at

**Insertion**

* GGA-CAA-TCA GCG-ACA-ATC-A

 proline -valine-serine arginine-cysteine-stop

**Deletion**

* GGA-CAA-TCA GGA-AAT-CA

proline -valine-serine proline-leucine

Example:

Cri du chat syndrome

* Due to a \_\_\_\_\_\_\_\_\_\_\_\_\_\_ of part of the short arm of chromosome 5
* Occurrence: 1/50,000 births
* Crying babies sound like cats; mental disability
* Death by about 4 years

**Chromosome Mutations**

**Karyotype** = chart of metaphase \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ pairs arranged according to length and location of the centromere

Used to pinpoint unusual chromosome \_\_\_\_\_\_\_\_\_\_\_\_\_\_ in cells

 Nondisjunction

During meiosis, the homologous chromosomes fail to \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ properly

This can result in two types of chromosomal \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_:

(a) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (have an extra set of chromosomes)

(b) \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (missing one set of chromosomes)

[**Edward’s syndrome (trisomy 18)**](http://www.trisomy.org/)**:**

**Occurs in 1:6000 or 1:8000 live births; very few survive birth.**