Bio12AP- **Alterations of Chromosome Number or Structure** Name: \_\_\_\_\_\_\_\_\_\_\_\_\_

Sec 15.4 **Cause Some Genetic Disorders** Date: \_\_\_\_\_\_\_\_\_\_\_\_\_\_

Pg 297 Block: \_\_\_

**Genetics Concept # 4**

**Learning Goal**: I can apply the theory of chromosomal inheritance to analyze changes in chromosome number and structure



**I. Alteration of Chromosome Number**

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

* Occurs when homologous chromosomes do not separate properly during meiosis II.

**Result**? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

* Incorrect number of a chromosome
	+ Trisomic: three copies of the chromosome
	+ Monosomic: one copy of the chromosome

 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

* More than two complete sets of chromosomes

**Other ways chromosome abnormalities happen…**

***Mitosis*:** problems with cell division

***Maternal Age*:** Women are born with all the eggs they will ever have. Errors may crop up in the eggs' genetic material with age. Because men produce new sperm throughout their lives, paternal age does not increase risk of chromosome abnormalities.

***Environment*:** No conclusive evidence however it is still possible that the environment may play a role in the occurrence of genetic errors.

**Most common non disjunctions in humans:**

Trisomy 21:

Trisomy 18:

Trisomy 13:

**Sex Chromosome Aneuploidy**

XXY-

XYY-

XXX-

**II. Alteration of Chromosome Structure pg 300**

Portions of a chromosomes may be \_\_\_\_\_\_or \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ during crossing over



**a) Deletion**: Fragment is lost.  Missing \_\_\_\_\_\_\_\_\_\_\_\_.

 *Example: Cri du Chat syndrome- deletion in chromosome 5 in humans,*

 *Symptoms: small head, cry sounds cat like, death in infancy*

**b) Duplication**: Extra fragment.  \_\_\_\_\_\_\_\_\_\_\_ genes.

**c) Inversion:** a chromosomal fragment reattaches in \_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**d) Translocation**: when a fragment of a chromosome becomes attached to a \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

*Example: Chronic Myelogenous leukemia (CML) &*



*the Philadelphia chromosome*

**Karyotypes:**

-are images that show the number, size & types of chromosomes

-used to diagnose some



genetic disorders

Karyotyping activity online at: <http://www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping.html>