## Unit 5: Mendelian Genetics Content Outline: Abnormal Chromosome Number (5.6) "When Meiosis Goes Wrong"

- I. Chromosomal Errors than can occur:
  - A. These could occur during Mitosis or Meiosis.
    - 1. They would occur during the Anaphase Stages where chromosomes are moving.
    - 2. They could also occur during Crossover where gene DNA segments are moving.
    - 3. Please make sure that student understand the difference between a gene mutation and a Chromosomal abnormality. This is a very important concept that tests love to ask questions on this comparison. Gene is associated with the *nucleotide sequence* (point and reading frame) where chromosomal are about *structure of chromosomes or numbers of chromosomes*.
  - B. Two *types* of errors can occur:
    - 1. Chromosomal Number (Aneuploidy means "Abnormal number of chromosomes" See Part II
      - below)
      - a. This is the result of **non-disjunction**. (*Failure to separate* during Anaphase.) Can you "see" a possible definition in the term?
      - b. **Trisomic** (*Three* of 1 *kind* of chromosome.)
      - c. Monosomic (*Missing one*, the other half of the pair.) (It is located in the **Trisomic** gamete.)
      - d. Please help students see the connection between the two terms. Students should understand that the chromosome just did not disappear as that would be against the 1<sup>st</sup> Law of Matter... Matter is neither created nor destroyed; just transformed or transferred.
      - e. **Polyploidy** (*Many extra sets* of chromosomes.)
        - i. 3n (triploid) Three "halfs" are in this cell.
        - ii. 4n (tetraploid) Four "halfs" are in this cell.
        - iii. Deadly in most animals; Plants not really affected.
      - 2. Individual Chromosome *Structure* (Please help students "see" the terms as well as the relationship between them in one exists.)
        - a. These occur because of faulty crossover.
        - b. **Deletion** Chromosome segment is "*missing*". It got *stuck on the other* homologous chromosome during crossover.
        - c. **Duplication** A chromosome segment was "*copied*" twice. (Two genes on one chromosome. It is "*missing*" from the other homologous chromosome.)
        - d. Inversion A chromosomal segment is "backwards". It was inverted backwards during crossover.
        - e. **Translocation** A chromosomal segment is *attached to a different autosome*. It *accidentally* broke loose and ended up on another chromosome.

## II. Syndrome

- A. This term refers to an organism "possessing" the identifying traits of a particular genetic disorder.
- B. Human Genetic Disorders due to two abnormal chromosomal number (#) or structure:
  - 1. **Down's Syndrome** ( # )
    - a. This affects about 1 in 700 births.
    - b. This individuals possess an *Extra 21 Autosome* (A.K.A. **Trisomy 21**)
    - c. General syndrome features (Please show a facial picture and discuss the characteristics.)
    - d. Mainly the result of women of advanced age having babies.
  - 2. Turners Syndrome (XO) ( # )
    - a. General characteristics: These individuals appear fairly normal. They just are lacking the "sexual" characteristics we normally see in individuals. The "sexual" traits are missing because there is <u>no</u> second sex chromosome to help create those traits. These individuals are usually raised as females.

Remember, there are <u>no</u> cures for <u>all</u> of these disorders; <u>only treatments</u> because the problem is genetic!