Genetic and Chromosomal Disorders
- an overview -

Things Can Go Wrong
With Genes and/or Chromosomes

I. DNA Based Disorders

A. Dysfunctional Genes
   1. a gene can be missing some of its nucleotides or some of the nucleotides present may be in the wrong order
      a. the resulting protein may not work at all, or work less than 100%, or in rare cases do something unexpected

B. Missing Genes
   1. a person can be born missing an entire gene or missing so much of a particular gene that in either case there is no gene product (protein) produced for that particular gene

C. Epigenetic Factors
   1. small molecules bonding to a gene in such a way as the gene turns off – this can change during a lifetime
II. Chromosomal Based Disorders

A. Non-disjunction

1. a tetrad (4 homologous chromosomes) at the end of meiosis I does not separate properly
   a. can produce viable gametes with extra chromosomes or…..
      1. in one special case, the embryo can survive with only one X chromosome (the Y chromosome is the only chromosome that can be missing) *and early development genes are on the X Chrom.
   b. when these gametes fuse with a normal gamete during fertilization, the resulting person will have an abnormal number of chromosomes
   c. approximately 1 birth in every 1000 is affected by an abnormality involving non-disjunction of the sex chromosomes

B. Health Issues

1. every non-disjunction occurrence greatly affects the individual’s health, life span, and/or mental capacity
   a. ethical considerations
      1. chromosomal disorders are easy to detect before birth
      2. parents and doctors are faced with issues that past generations NEVER had to face.
      3. how should a parent react to news that their child will be born with a non-disjunction condition?
The End