# **Additional Bio Notes**

## **Recessive Disorders**

- Cystic fibrosis build up of mucous on certain cells, typically die young
- Tay-Sachs disease dysfunctional enzyme causes brain malfunction, die young
- Sickle-cell disease substitution of a single amino acid which alters the shape of hemoglobin

# **Dominant Disorders**

- Huntington's Disease degenerative disease of the nervous system, die late 35 to 40
- Achondroplasia dwarfism

#### **Chromosomal Basis for Sex**

- XX and XY humans
- XO and XX males only have one sex chromosome
- ZZ and ZW males have same sex chromosome vs. females who have two different (reverse of humans)
- Haploid and Diploid males develop from unfertilized eggs (they have no father)
  <u>Sex-Linked Disorders</u>
- Color blindness can't see colors
- Duchenne muscular dystrophy weakening of muscles and loss of coordination
- Hemophilia absence of a protein required for clotting

#### X-Inactivation in females

- Barr body only one X is completely active in females the other condenses into a Barr body
- Reactivated in meiosis

# **Alteration of Chromosome Number and Genetic Disorders**

- Nondisjunction homologous chromosomes don't separate during meiosis
- Aneuploidy abnormal chromosome number
  - Trisomic -2n + 1
  - Trisomy 21 Down syndrome
  - Monosomic 2n 1
- Polyploidy 3n or 4n

## **Alteration of Chromosome Structure**

- Deletion loss of a chromosomal fragment
- Duplication fragment joins another chromosome
- Inversion deletion rejoins original chromosome in reverse order
- Translocation deletion joins a non-homologous chromosome