Chapter 12 and 13 / Genetics

I. Introduction to Genetics--study of inheritance
   A. Traits are inherited
      1. passed from one generation to another
      2. information is carried along on genes--regions of DNA
   B. Describing an organism’s genetic makeup
      1. phenotype = visible trait; expression of the genes
      2. genotype = actual codes or genes on DNA
   C. Why does variation exist?  How is it determined?

II. Mendelian Genetics
    Gregor Mendel  1866

   A. Early genetic observations
      1. forms of traits appear in certain offspring of a specific cross
      2. certain forms will be more highly represented than others (dominate)
      3. some forms of a trait will not show up in one generation, but may reappear unchanged in the following generation
   B. Genetic experiments with pea plants (Pisum sativum)
      1. easy to grow, reproduce quickly
      2. easily recognizable traits
      3. have enclosed flowers that can “self-fertilize”
   C. Mendel’s experiments
      1. started out with pea plants of “pure” strains
      2. made experimental crosses between different varieties
         --looked at one trait at a time: monohybrid cross
         --obtained hybrids in F1 generation
3. let hybrid’s offspring self-fertilize for several generations
   (F2, F3, etc. generations)

   --counted number of offspring of each type of trait for the succeeding generations

D. Mendel’s results

1. F1 generation of purple x white cross = all purple flowers

   purple = dominant trait

   white = recessive trait

2. F2 generation
   some offspring had recessive (white) trait
   proportions of purple to white = 3/4 to 1/4 (3:1)

3. subsequent generations (self-fertilize)

   purple: 2/3 mixture, 1/3 always purple
   white: always white

Mendelian ratio 3/4 dominant to 1/4 recessive in F2 generation of pure cross

F2 generation really 1:2:1 genotypic ratio
   --Mendel described phenotypes

E. Interpretation of results-- genotype frequencies

1. WW x ww    2 alleles of the gene

2. heterozygous vs. homozygous

3. Punnet square to show genotype outcome

    in a homozygous dominant x homzygous recessive cross, all offspring heterozygous

III. Mendel’s Laws

A. First Law: Law of Segregation
   --refers to Mendel’s observed ratios
   --alternative forms of a trait are distinct (no blending)
   --in heterozygous individuals, gametes have equal probability of possessing either allele
B. 2nd Law: Law of Independent Assortment  
--based upon experiments with 2 traits at a time:  
**dihybrid cross**  

Law states: alleles for different genes sort independently during meiosis  

Note: crossing over during meiosis → genetic recombination  

--Application: see if two traits are related (linked) or independent

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**Chromosomes and Exceptions to Mendelian Genetics**

IV. Sex Determination and Associated Traits  
A. Sex chromosomes  

X and Y  

B. Sex-linked traits (X-linked traits)  
--associated with X chromosome, not Y  

Therefore, recessive traits tend to be expressed more in males.  
  e.g., hemophilia; red-green color blindness  

C. X-inactivation  
In early development, one X chromosome randomly “shut off” per cell  
  e.g., calico cats  

V. Other Exceptions to Mendelian Genetics  
A. Incomplete dominance  
  heterozygote has phenotype intermediate between homozygotes  

B. Codominance  
If heterozygous, more than one allele expressed at a time  
  e.g., ABO blood groups  

C. Polygenes  
  alleles at different loci influence a single trait  
  e.g., skin color in humans – additive effect  

D. Lethal Alleles  
  e.g., recessive alleles that cause death of the embryo or fetus
VI. Chromosome Structure
   A. Chromosomes have particular sizes and shapes influenced by centromere position

   B. Assessing normal chromosome number and structure karyotype

   C. Abnormal chromosome structure e.g., missing or additional parts of chromosomes inverted (flipped) or translocated parts

VII. Abnormal Chromosome Number
   A. Polyploidy extra complete sets of chromosomes

   B. Aneuploidy extra or missing individual chromosomes

      1. Down syndrome: trisomy 21
      2. Turner’s syndrome: XO
      3. Triplo-X: XXX
      4. Klinefelter’s syndrome: XXY
      5. XYY

VII. Some Genetically Transmitted Diseases
   Phenylketonuria (PKU)

   Cystic fibrosis

   Parkinson’s Disease

   Sickle-cell Anemia

   Tay-Sachs Disease

   Huntington’s Disease

   Hemophilia