CSC 314, Bioinformatics Lab #1: OMIN and Genetics **Name:_____**

Online Mendelian Inheritance in Man (OMIM) is an online catalogue of human genes and genetic disorders (http://www.omim.org).

In Parts I-III of this lab, you will use the OMIM database along with your knowledge of genetics to answer questions about several genetic diseases. We will look at ID# 306700 as an example. In Part IV, you will apply your knowledge of genetics to answer questions about several genetic crosses.

Part I, genetic susceptibility to breast cancer -- Pull up the entry with ID# 612555.

1. What gene is this entry for?
2. What chromosome is the gene located on?
3. For heterozygous females, what is the lifetime risk of breast cancer?
4. Is this phenotype *dominant* or *recessive*?
5. Suppose that a heterozygous female mates with a male who does not have an increased susceptibility of developing breast cancer (i.e., is homozygous recessive). What is the probability that their child has an increased susceptibility to breast cancer? (Note: you must construct and show an appropriate Punnett square to answer this problem).

Part II, color-blindness -- Pull up the entry with ID# 303800

1. What gene entry is this for?
2. What chromosome is the gene located on?
3. Is this phenotype *dominant* or *recessive*? (Hint: look under the *Inheritance* section of the main page)
4. Suppose that a female carrier mates with a male who is *not* colorblind as a result of this gene. Construct a Punnett square to answer the following questions:
    a. What is the probability that a male child produced from this union is colorblind?
    b. What is the probability that a female child produced from this union is colorblind?

Part III, Parkinson's disease -- Pull up the entry with ID# 612953

1. What gene entry is this for?
2. What chromosome is the gene located on?
3. Is this phenotype *dominant* or *recessive*?
4. Suppose that a female carrier mates with a male carrier (**Hint:** If a gene is *not* sex-linked, then you do should *not* keep track of the sex chromosomes; the phenotypic ratios will be the same for males and females). Construct a Punnett square to answer the following questions:
    a. What is the probability that a male child produced from this union will develop Parkinson disease-14?
    b. What is the probability that a female child produced from this union will develop Parkinson disease-14?

Part IV, Additional Genetics Questions

1. *Incomplete dominance*. A dog with a black coat mates with a dog with a white coat, and all of the offspring have gray coats, a result due to incomplete dominance. What must the genotypes of the parents and the offspring be, assuming that *B* = black coat and *b* = white coat?

2. *Codominance in blood types*.

    a. If a male with type *A* blood (with genotype I^Ai) mates with a female having type *B* blood (with genotype I^BI^B), what are the phenotypic ratios for the blood types of the offspring?

    b. If an individual has type *O* blood, which of the following is NOT possible, and **why**?
        i. At least one parent has type *A* blood
        ii. At least one parent has type *B* blood
        iii. At least one parent has type *AB* blood
        iv. At least one parent has type *O* blood

3. *Genetic cross with independent assortment (unlinked traits)*. Wolves are sometimes observed to have black coats and blue eyes. Assume that these traits are each determined by single genes that are located on *different* chromosomes. Also assume that normal gray coat color (G) is dominant to black (g) and brown eyes (B) are dominant to blue (b). Suppose that each individual has a normal-colored coat and brown eyes, and that both individuals are heterozygous for both traits. If these individuals mate, what are the expected phenotypic ratios of the offspring? Construct the appropriate Punnett square and answer the questions below:

    a) From your Punnett square, how many individuals are dihybrids?

    b) How many individuals are homozygous recessive for coat color and heterozygous for eye color?

    c) Fill in the blanks to state the expected phenotypic ratio of the offspring:

    ___ Gray coat, brown eyes : ___Gray coat, blue eyes : ___ Black coat, brown eyes : ____ Black coat, blue eyes

4. *Genetic cross with <u>dependent</u> assortment (linked traits)*. Repeat the previous problem but assume that the genes are located very close to each other on the *same* chromosome, and are therefore always *linked* (passed on together). Assume that in these individuals, each chromosome contains the dominant allele for one gene and the recessive allele for the other gene. Note: this is different than the independent vs. dependent assortment example in your notes. Construct a Punnett square, and indicate the phenotypic and genotypic ratio in this case:

   a) From your Punnett square, how many individuals are dihybrids?

   b) How many individuals are homozygous recessive for coat color and heterozygous for eye color?

   c) Fill in the blanks to state the expected phenotypic ratio of the offspring:

   ___ Gray coat, brown eyes : ___Gray coat, blue eyes : ___ Black coat, brown eyes : ____ Black coat, blue eyes