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

**Spring 2022**

***Note: This assignment must be submitted as a hardcopy in any format (handwritten or typed) at the beginning of class on the due date.***

*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.

**Note:** You must use proper notation for full credit, and must show Punnett squares, unless noted otherwise. When specifying a genotype, you should use a capital letter for a dominant allele and a lowercase letter for a recessive allele, and these should be defined (e.g., N = *normal* and *n* = hemophilia). For X-linked traits, your genotypes must indicate that the gene is on the X chromosome, e.g. XA, and that the gene is not on the Y chromosome, e.g., a male with the dominant phenotype would be XAY. In addition, you should only consider the X and Y chromosomes for sex-linked traits. If a trait is not sex-linked, then the phenotypes apply equally to males and females.

Example, Hemophilia A – Pull up the entry with ID #306700

1. What gene is associated with Hemophilia in this entry?
2. What chromosome is the gene on?
3. Look at the Clinical Synopsis, under “Miscellaneous”, to find the main phenotype associated with this disease (this is the second one listed).
4. What is the inheritance of this gene and phenotype?
5. Suppose that a female carrier mates with a male who is healthy (does not have hemophilia).
   1. What is the probability that a female child has hemophilia?
   2. What is the probability that a male child has hemophilia?

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

1. What gene is associated with breast cancer in this entry?
2. What chromosome is the gene located on?
3. Based on the Clinical Synopsis, what is the lifetime risk of breast cancer in heterozygous females (i.e., mutation carriers)?
4. Is this phenotype *dominant* or *recessive*?
5. Suppose that a female with the mutation (i.e., a heterozygous female) mates with a healthy male (the male’s genotype is homozygous recessive). What is the probability that their child has an increased susceptibility to breast cancer?

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

1. What gene is associated with color-blindness in this entry?
2. What chromosome is the gene located on?
3. Is this phenotype *dominant* or *recessive*? (Hint: look at the first paragraph under the *Inheritance* section of the main page)
4. Suppose that a female who is colorblind mates with a male who is colorblind. For (a) and (b), make sure to define the alleles.
   1. What is the genotype of the female?
   2. What is the genotype of the male?
   3. What is the probability that a male child produced from this union is colorblind?
   4. 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 is associated with Parkinson’s disease in this entrry?
2. What chromosome is the gene located on?
3. Is this phenotype *dominant* or *recessive*?
4. Suppose that a heterozygous female mates with a heterozygous male. What is the probability that a 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. Assume that C*B* = black coat and CW = white coat?
   1. What are the genotypes of the parents?
   2. What are the genotypes of the offspring?

1. *Codominance in blood types*.
   1. If a male with type *A* blood (with genotype *IAi*) mates with a female having type *B*

blood (with genotype *IBi*), what are the phenotypic ratios for the blood types of the offspring?

\_\_\_ A : \_\_\_ B : \_\_\_ AB : \_\_\_\_ O

* 1. Suppose that one parent has type AB blood, and the blood type of the other parent is not known.
     1. If a child has Type A blood, what are the possible genotypes of the other parent?
     2. Can the child have Type O blood? If so, how? If not, why not?

1. *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 parent has a normal-colored coat and brown eyes, and that both individuals are heterozygous for both traits. Use the Punnett Square below to answer the following questions:

**GgBb x GgBb**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **GB** | **Gb** | **gB** | **gb** |
| **GB** | GGBB | GGBb | GgBB | GgBb |
| **Gb** | GGBb | GGbb | GgBb | Ggbb |
| **gB** | GgBB | GgBb | ggBB | ggBb |
| **gb** | GgBb | Ggbb | ggBb | ggbb |

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

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

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

1. *Genetic cross with dependent 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 similar to the dependent assortment example that we went over in class, but the linkage is different. Construct a 2x2 Punnett square, and answer the questions below:

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

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

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