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

**Spring 2025**

***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 phenotypic ratios are the same for 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 mutation carriers (i.e., individuals who are heterozygous)? Note: these statistics are for females, but male carriers also have an increased risk of breast cancer.
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? Note: sex here is not relevant, since the gene is not sex-linked.

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 carrier mates with a male who is not 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, Charcot-Marie-Tooth disease -- Pull up the entry with ID #607706

Note: this condition is named after the people who discovered it; it has nothing to do with your teeth.

1. What gene is associated with Charcot-Marie-Tooth disease in this entry?
2. What chromosome is this gene on?
3. Is this phenotype *dominant* or *recessive*?
4. What is the neurologic impact on a person’s distal limbs (lower arms and legs)?
5. Suppose that two heterozygous individuals have a child. What is the probability that the child will inherit this disease?
6. The previous question (5) describes a *monohybrid* cross. Why does the term *monohybrid* refer to in this case?

Part IV, Additional Genetics Questions

1. *Incomplete dominance*. Assume that coat color in a dog is determined by incomplete dominance with C*B* = black and CW = white, and that dogs that are heterozygous have gray coats. If a dog with a grey coat mates with another dog with a gray coat, what is the phenotypic ratio of coat color in the offspring?

\_\_\_\_ black : \_\_\_\_ gray : \_\_\_\_ white

1. *Codominance in blood types*.
   1. If a male with type *AB* blood (with genotype *IAIB*) 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 a child has Type O blood, and one parent has type O blood. For each blood type, specify the possible genotypes for the other parent, or write “not possible” if it is not possible that the parent has the given blood type.

Type A:

Type B:

Type AB:

Type O:

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. The chromosomes of both parents are shown below. Construct a 2x2 Punnett square, and answer the questions below.

x

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