Population Genetics
Problem Set 2 Key

1. Silic located data on MN blood type frequencies from a sample of 429 Navaho Native Americans in New Mexico. The phenotypic results were:

M	363
MN	57
N	9

a) What is the frequency of the M allele?
b) What is the frequency of the N allele?

1. n = 429   MM = 363   MN = 57   NN = 9   p = M   q = N

a. $p = \frac{363 + ½(57)}{429}$ = **.913 ~ .91**

b. q = 1 – p = **.087 ~ .09**   or $q = \frac{9 + ½(57)}{429}$ = **.087 ~ .09**

2. Boyd (1950) tested and reported the **MN** blood type frequencies for a sample of 361 Navaho Native Americans in New Mexico. The phenotypic results were:

305 **M**
52 **MN**
4 **N**

a. What is the frequency of the **M** allele?
b. What is the frequency of the **N** allele?
c. What are the expected genotypic frequencies under Hardy-Weinberg equilibrium?
d. Use chi-square value (χ^2) to test whether the observed genotypic frequencies are consistent with Hardy-Weinberg equilibrium.

p = M = [305 + (1/2)52] / 361 = **.9169**

q = N = [(1/2)52 + 4] / 361 = **.0831**

$p^2 + 2pq + q^2$	<u>actual</u>
MM = $p^2 = (.9169)^2$ = **.8407**	305/361 = .8449
MN = 2pq = 2(.9169)(.0831) = **.1524**	52/361 = .1440
NN = $q^2 = (.0831)^2$ = **.0069**	4/361 = .0111

$\chi 2 = \Sigma\ [(\text{Expected} – \text{Obtained})^2 / \text{Expected}]$

$[(303.49 – 305)^2 / 303.49] + [(55.02 – 52)^2 / 55.02] + [(2.49 – 4)^2 / 2.49] = .0075 + .1658 + .9157 = 1.089$

Degrees of freedom = n – (r alleles) = 3-2 = 1
$X^2_{1df} = 3.84$ **1.089< 3.84**
Fail to reject the hypothesis – so consistent with H-W

3. A survey indicated that one person in 39,000 had a particular genetic recessive disorder.
   a. What is the frequency of the recessive allele?
   b. Approximately how many of these people are heterozygous for the disorder?

   a. q_{aa} = 1/39,000 q_a = sqrt(1/39,000) = **.0051**

   b. q_A = 1 - .0051 = .9949 $2q_Aq_a$ = 2(.9949)(.0051) = .0101
   39,000 * .0101 = 393.9 people ~ **394 people**

4. Assume that a sex-linked recessive trait is observed in 25% of all the males and that the population is in Hardy-Weinberg equilibrium.
   a. What is the frequency of the recessive allele?
   b. What is the frequency of the dominant allele?
   c. What are the expected genotypic frequencies in the female?

   Ay = .75 ay = .25
   a. q_a = **.25**

   b. p_A = **.75**

   c. AA = (.75)2 = **.5625**
   Aa = 2(.75)(.25) = **.375**
   Aa = (.25)2 = **.0625**

5. Four babies were born in a hospital on the same night, and their blood groups were later found to be **O, A, B** and **AB**. The four pairs of parents were:

   O and **O**
   AB and **O**
   A and **B**
   B and **B**

   Assign the four babies to their correct parents.

Baby	Parent
O	O,O
A	AB,O
B	B,B
AB	A,B

6. The frequency of **ABO** types in a group of about 300 samples was 81 percent **O**, 19 percent **A**, and no **B** or **AB**. Estimate the frequencies of the **A, B**, and **O** alleles in this population.

Genotypic frequencies: .81 O .19 A 0B 0AB
$q_O = \sqrt{(.81)}$ = **.9**
q_A = 1 - .9 = **.1**
q_B = **0**

Remember not to take the square root of .19 because A blood type could be AA or AO whereas O can only be OO.

7. The following table lists the frequencies for the blood group phenotypes observed in 190,177 persons in Great Britain (Dobson and Ikin, 1946).

Phenotype	Observed Frequency
A	79,334
B	16,280
AB	5,781
O	88,782
Total	190,177

What are the six genotypic frequencies under the condition that the population of **A,B,O** blood types are following Hardy-Weinberg equilibrium?

	Observed		Expected
A	79,334	P_{A-}	$P_A^2 + 2P_AP_O$
B	16,280	P_{B-}	$P_B^2 + 2P_BP_O$
AB	5,781	P_{AB}	$2P_AP_B$
O	88,782	P_{OO}	P_O^2
	190,177		

$P_O = \sqrt{(88{,}782/190{,}177)} = .6833$
$P_A = \sqrt{(P_{A-} + P_{OO})} - \sqrt{(P_{OO})}$
$= \sqrt{(79{,}334/190{,}177 + 88{,}782/190{,}177)} - \sqrt{(88{,}782/190{,}177)} = .2569$
$P_B = \sqrt{(P_{B-} + P_{OO})} - \sqrt{(P_{OO})}$
$= \sqrt{(16{,}280/190{,}177 + 88{,}782/190{,}177)} - \sqrt{(88{,}782/190{,}177)} = .060$

$P_O + P_A + P_B = .6833 + .2569 + .060 = 1.0002$

$W = 1 – 1.0002 = -.0002$
$P_O' = (P_O – 1/2W)(1 – 1/2W) = (.6833 – ½(-.0002))(1 – ½(-.0002)) = .6835$
$P_A' = (P_A + W)(1 – 1/2W) = (.2569 + -.0002)(1 – ½(-.0002)) = .2567$
$P_B' = (P_B + W)(1 – 1/2W) = (.060 + -.0002)(1 – ½(-.0002)) = .0598$

$P_O' + P_A' + P_B' = .6835 + .2567 + .0598 = 1$

$AA = (.2567)^2 = .0659$
$AO = 2(.2567)(.6835) = .3509$
$BB = (.0598)^2 = .0036$
$BO = 2(.0598)(.6835) = .0817$
$AB = 2(.2567)(.0598) = .0307$
$OO = (.6835)^2 = \underline{.4672}$
1

8. Xeroderma pigmentosum is a severe disease in which the affected individual is not able to repair the damage done to skin and other tissue by ultraviolet radiation. This disease is known to be autosomal recessive and is found in approximately 1 in 333335 people. What is

the expected frequency of heterozygous people? (i.e. What is the expected frequency of carriers?)

P_{rr} = 1/333,335

$P_r = \sqrt{P_{rr}} = \sqrt{(1/333,335)}$ = **.00173**
P_R = 1 - .00173 = **.99827**

$2pq = 2P_RP_r$ = 2(.00173)(.99827) = **~.0034**

9. Allele A is dominant to a and allele B is dominant to b. A plant was a self-fertilizing plant, and heterozygous for both pairs of alleles.
a) What is the probability that all of 8 offspring would have the A_B_ phenotype?
b) What is the probability that the first offspring will be identical to the parent?
c) What is the probability that the first offspring is homozygous at both loci? Please note that more than one genotype is homozygous.

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

a. A_B_ = 9/16
$(9/16)^8$ = **.01**

b. P(AaBb) = (4/16) = **1/4**

c. P(AABB + AAbb + aaBB + aabb) = (4/16) = **1/4**

10. In a population of Holsteins, an autosomal recessive allele, A_2, is found at a frequency of $Pr(A_2) = 0.34$. A sample of the population found 39 individuals with no sign of the condition, while 11 individuals were affected.
a) Among the total population of Holsteins, what proportion are expected to be heterozygous?
b) From the sample alone, how many individuals in the sample are expected to be heterozygous?
c) Among the total population of Holsteins, what proportion are expected to be homozygous for A_1 and what proportion are expected to be homozygous for A_2?
d) From the sample, how many individuals are expected to be homozygous for A_1?
e) Is the sample representative of the total population? Use chi-square to test hypothesis. (Hint: You only have observations on two groups, affected and unaffected.)

P_{A2} = .34 P_{A1} = 1-.34 = .66 39A_1_ 11A_2A_2

a. 2pq = 2(.34)(.66) = **.45**

b. $A_2 = \sqrt{11/50}$ = .4690

$2pqN = 2(.469)(.531)(50) =$ **24.9 Holsteins**

c. $P_{A1A1} = (.66)^2 =$ **.4356**
$P_{A2A2} = (.34)^2 =$ **.1156**

d. $P_{11}N = (.531)^2 * 50 =$ **14**

e. $(.1156)(5) = 5.78$
$50 – 5.78 = 44.22$

$X^2_{calc} = \sum[(\text{Expected} – \text{Observed})^2 / \text{Expected}]$
$X^2_{calc} = [(5.78 – 11)^2 / 5.78] + [(44.22 – 39)^2 / 44.22]$
$= 4.71426 + .616201 = 5.33$

$X^2_{table} = 3.84$ (1df, $\alpha = .05$)

$X^2_{calc} > X^2_{table}$ 5.33 > 3.84, so reject the hypothesis that the population is in H-W
*With an alpha probability of 0.05 of type one error rate, I reject the hypothesis that the population of Holsteins is in Hardy-Weinberg equilibrium. (Note: it is possible to do run a test for a dominant trait in this case since the population allele frequencies are provided rather than calculated from the data, which adds one degree of freedom.)

11. The MN blood group was tested in nomads of the Sahara Desert. A sample of 1842 people were tested for the presence of the M and N antigens. Results were 1098 people were MM, 575 people were MN and 169 people were NN.
a) What are the M and N allele frequencies?
b) Use chi-square to evaluate if the population was in Hardy-Weinberg equilibrium.

1098/1842 MM 575/1842 MN 169/1842 NN

a. $P_M = \frac{1098 + ½(575)}{1842} =$ **.752 ~ .75**
$P_N = \frac{169 + ½(575)}{1842} =$ **.248 ~ .25**

b. $(.75)^2(1842) = 1036$
$2(.75)(.25)(1842) = 691$
$(.25)^2(1842) = 115$

$X^2_{calc} = \sum[(\text{Expected} – \text{Observed})^2 / \text{Expected}]$
$X^2_{calc} = [(1036 – 1098)^2 / 1036] + [(691 – 575)^2 / 691] + [(115 – 169)^2 / 115]$
$= 3.710 + 19.473 + 25.357 = 48.54$

$X^2_{table} = 3.84$ (1df, $\alpha = .05$)

$X^2_{calc} > X^2_{table}$ 48.54 > 5.991, so reject the hypothesis that the population is in H-W
*With an alpha probability of 0.05 of type one error rate, I reject the hypothesis that the population of nomads is in Hardy-Weinberg equilibrium, and conclude they are not in equilibrium.