

Session 1 of Computer practical Work

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1. Estimation of genotype and allele frequencies from a genotyping survey

Both scripts for this exercise (mandatory one and optional can be found in the file with the scripts).

2. Random mating in a mendelian population and HWE

Ex set 1 (Modify the previous script to compute the impact of assortative mating on the genotype and allele frequencies):

The original script and the corresponding modifications can be found in the file with scripts.

Ex set 2: (i did not manage to do none of the 2 scripts for this set of ex, sorry)

- Segregation:

In this case, knowing that the segregation favours A, so it is logical to expect more A_ in the next generations

- X-linked

Ex set 3:

1. Two populations start with the following genotypic frequencies:
 - Population I 0.24AA 0.32Aa 0.44aa
 - Population II 0.33AA 0.14Aa 0.53aa.
 - For population 1: AA - 0.16, Aa - 0.48, aa - 0.36
 - For population 2: AA - 0.16, Aa - 0.48, aa - 0.36
2. In a population that is in Hardy-Weinberg equilibrium, the frequency of allele A is 0.7 and that of allele a is 0.3. What will be the allele and genotype frequencies after a generation, if only the heterozygotes are crossed?
 - After deducing the values from the statement we get that the allele frequencies in the next generation are: p - 0.699 and q - 0.301 and genotypic frequencies are : AA - 0.48, Aa - 0.42, aa - 0.1
3. Phenylketonuria (PKU) has an autosomal recessive inheritance. In a human population, a person with PKU (homozygous recessive) was found for every 10,000

individuals analyzed. Assuming that this population is in HWE for this locus, what are the allele frequencies of that locus?

- $p = 0.99, q = 0.01$
- 4. If 4% of individuals in a population that is in HWE manifest the recessive phenotype, what is the probability that a descendant of two individuals who do not express this recessive character, do express it?
 - $(\text{Probability of picking a heterozygous})^2 \times (\text{probability of getting a recessive homozygous}) = 0.077^2 \times 0.25 = 0.00333$ or 33.3%
- 5. In the human species, a genetic disease is determined by an autosomal dominant gene. In a population the percentage of people affected by the disease is 19%. If the population is in Hardy-Weinberg equilibrium, what is the probability that a randomly chosen couple are both affected and has a normal child?
 - $(\text{Probability of picking a heterozygous})^2 \times (\text{probability of getting a recessive homozygous}) = 0.18^2 \times 0.25 = 0.0081$ or 8.1%
- 6. 1 in 1700 US Caucasian newborns have cystic fibrosis. C is the normal allele, dominant over the recessive c. Individuals must be homozygous for the recessive allele to have the disease.
 - $q^2 = 0.00059, q = 0.02425, p = 0.97575, 2pq = 0.04732$ and assuming HWE a 6 newborns would have cystic fibrosis
- 7. If 60% of men in a population that is in HWE have a dominant character linked to the X, what percentage of women are expected to present this character?
 - In this case, the % of females that will present the character is the sum of dominant homozygous and heterozygous, which are: $0.48 + 0.36 = 84\%$
- 8. A recessive X-linked disease occurs in 1% of men. What is the frequency of the allele of the disease if we assume that the population is in HWE?
 - Assuming the same frequency of females and males in the population and that the population is in HWE, the frequency is going to be same, 0.01.

3. HWE chi-square test on counts from a genotyping survey

The script for this exercise can be found in the script file.

1. 361 Navajo individuals were analyzed in New Mexico for the MN locus: 305 M types, 52 MN and 4 N:

- a) By using the script to test if the data is in HWE and having a p-value of 0.4 meaning that it is pretty far away from HWE.
 - b) 0.01
 - c) 0.14
- 2. What allelic frequency will generate twice as many recessive homozygotes as heterozygotes?
 - The allelic frequency to generate double homozygotes as heterozygotes can be achieved by using $p = 0.2$ and $q = 0.8$
- 3. Three alleles A, B, and C of the red cell acid phosphatase locus were found in a sample of 178 English Individuals.
 - a) allelic frequencies are: A - 0.35, B - 0.6 and C - 0.05. By performing a chi-square test we can see that this dataset is not consistent with HWE
- 4. I did not manage to figure out how to solve this problem

4. Estimation of allele and genotype frequencies from counts of dominant and recessive phenotypes

The script for this exercise can be found in the file with the scripts

- 1. What is the frequency of heterozygotes Aa in a population with random mating in which the frequency of the dominant phenotype is 0.19?
 - $A_+ = 0.18$
- 2. 2. What is the frequency of heterozygotes Aa in a population with random mating if the frequency of the recessive phenotype aa is 0.09?
 - $A_+ = 0.42$
- 3. In the human population, one in every 10,000 babies is albino. Calculate the approximate frequency of the allele produced by albinism, knowing that it is autosomal and recessive
 - 0.009
- 4. What proportion of non-albino individuals are carriers?
 - 0.019

5. If 9% of an African population is born with a severe form of sickle-cell anemia (ss), what percentage of the population will be more resistant to malaria because they are heterozygous (Ss) for the sickle-cell gene?
 - 0.42 or 42% of the population of Africa
6. A very large population of randomly-mating laboratory mice contains 35% white mice. White coloring is caused by the double recessive genotype, "aa". Calculate allelic and genotypic frequencies for this population.
 - $p = 0.41$, $q = 0.59$ and $AA = 0.17$, $Aa = 0.48$, $aa = 0.35$
7. After graduation, you and 19 of your closest friends (lets say 10 males and 10 females) charter a plane to go on a round-the-world tour. Unfortunately, you all crash land (safely) on a deserted island. No one finds you and you start a new population totally isolated from the rest of the world. Two of your friends carry (i.e. are heterozygous for) the recessive cystic fibrosis allele (c). Assuming that the frequency of this allele does not change as the population grows, what will be the incidence of cystic fibrosis on your island?
 - 0.25 or 25 of the population of the island
8. Suppose that premature baldness in man is determined by a single gene, B, which is dominant in males and recessive in females. If a human population in equilibrium presents 51% of bald men:
 - a) What is the frequency of bald women?
 - b) Assuming random mating, what proportion of random mating would occur between a bald man and a normal woman?
 - c) In what proportion of the marriages would the first child present baldness?
 - d) If a normal woman has a male child, what is the probability that he will become bald?
 - e) A bald woman has a daughter, but nothing is known about the father. What is the probability that this girl will become bald with time?
 - a) 8% , b) 20%, c) 10%, d) 25.5% , e) 25.5%