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POPULATION GENETICS AND MICROEVOLUTIONARY THEORY
MIDTERM EXAMINATION

Write your name on every page!

The following table may be useful in answering one or more of the questions. Use it as needed:

Probability	Chi-Square Test Statistic Value			
	df = 1	df = 2	df = 3	df=4
0.10	2.706	4.605	6.251	7.779
0.05	3.841	5.991	7.815	9.488
0.01	6.635	9.210	11.345	13.277

1. Briefly define (5 points each):

a) Proximate causation

A necessary and sufficient explanation for a phenomenon under study based on the content of the system in which the phenomemon is manifested.

b) disassortative mating

The preferential mating of individuals with unlike phenotypes above the expectations of random mating.

c) An evolutionary force

A process that can alter allele or gamete frequencies in the gene pool.

2. A sample of individuals is surveyed at two autosomal loci each with two alleles as follows:

Genotype	Locus 1	AA	Aa	aa	Total	Locus 2	BB	Bb	bb	Total
Number		22	36	42	100		40	40	20	100

a. (2 points) What are the allele frequencies for the A and B loci?

$$p_A = (44+36)/200 = 0.40$$

$$p_B = (80+40)/200 = 0.60$$

b. (4 points) What are the expected two locus genotype frequencies at Hardy-Weinberg equilibrium?

Multiply the single locus HW genotype frequencies to get the two locus equilibrium:

$$\mathbf{AABB, (.4)^2(0.6)^2 = 0.0576}$$

$$\mathbf{AABb, (.4)^2 2(0.6)(0.4) = 0.0768}$$

$$\mathbf{AAbb, (.4)^2 (.4)^2 = 0.0256}$$

$$\mathbf{AaBB, 2(0.4)(0.6) (.6)^2 = 0.1728}$$

$$\mathbf{AaBb, 2(0.4)(0.6) 2(0.6)(0.4) = 0.2304}$$

$$\mathbf{Aabb, 2(0.4)(0.6) (.4)^2 = 0.0768}$$

$$\mathbf{aaBB, (0.6)^2(0.6)^2 = 0.1296}$$

$$\mathbf{aaBb, (0.6)^2 2(0.6)(0.4) = 0.1728}$$

$$\mathbf{aabb, (0.6)^2 (.4)^2 = 0.0576}$$

c. (4 points) Given that $D=0.1$, what are the two locus gamete frequencies in the surveyed population?

$$\text{Use } D = g_{AB} - p_A p_B \text{ so } g_{AB} = (0.4)(0.6)+0.1 = 0.34; \text{ similarly, } g_{ab} = (0.6)(0.4)+0.1 = 0.34$$

$$\text{and } g_{Ab} = (0.4)(0.4)-0.1 = 0.06; g_{aB} = (0.6)(0.6)-0.1 = 0.26$$

d. (2 points) What is the standardized disequilibrium, D' , for the population described in part c?

$$\text{Use } D' = \frac{D}{\min(p_A p_b, p_a p_B)}, D > 0, \text{ so } D' = 0.1/\min[(0.4)(0.4), (0.6)(0.6)] = 0.1/0.16 = 0.625$$

3. The disequilibrium between two loci is $D = 0.3$ and the recombination frequency between them is 0.05.
a. (2 points) Assuming random mating, what is the D in the next generation?

Use $D_t = D_0(1-r)^t$, so $D_1 = 0.3(1-0.05) = 0.285$

- b. (2 points) Assuming both loci influence a trait associated with disassortative mating, do you expect the D in the next generation to be greater than, equal to, or less than the D calculated in part a)?

Disassortative mating causes D to decay at a faster rate than random mating, so D in this case should be less than 0.285.

- c. (2 points) Assuming both loci influence a trait associated with assortative mating, do you expect the D in the next generation to be greater than, equal to, or less than the initial D ?

Assortative mating causes D to increase, so D in this case should be greater than 0.3.

- d. (2 points) Assuming the population has a system of mating with $f=0.2$, do you expect the D in the next generation to be greater than, equal to, or less than the D calculated in part a)?

Inbreeding causes D to decay at a slower rate than random mating, so D in this case should be greater than 0.285.

- e) (2 points) Assuming the population has a system of mating with $f=-0.2$, do you expect the D in the next generation to be greater than, equal to, or less than the D calculated in part a)?

Avoidance of inbreeding causes D to decay at a faster rate than random mating, so D in this case should be less than 0.285.

4. 10 isolated demes are drawn at random from zygotes produced by 5,000 unrelated adults mating at random for a locus with two alleles, with each allele having a frequency of 0.5 in the adult population. Assume that all 10 demes are of equal size (both census and effective in all senses), have the same system of mating, and genetic drift is the only evolutionary force operating. All individuals in the 10 isolated demes are surveyed genetically and the following allele frequencies are found: 0.1, 0.2, 0.3, 0.4, 0.5, 0.5, 0.6, 0.7, 0.8, 0.9. Use the population of 5,000 unrelated adults as the reference generation.

a. (2 points). What is the inbreeding effective size of each deme?

Inbreeding effective size is a function of the number of parents, which is 5,000.

b. (6 points) What is the variance effective size of each deme?

First, calculate the variance of the allele frequencies (given the mean of 0.5)

Variance =

$$[(0.1-0.5)^2+(0.2-0.5)^2+(0.3-0.5)^2+(0.4-0.5)^2+2(0.5-0.5)^2+(0.6-0.5)^2+(0.7-0.5)^2+(0.8-0.5)^2+(0.9-0.5)^2]/10 = 0.060 \quad (3 \text{ points})$$

Then use $N_{ev} = \frac{1}{2 \left[1 - \left(1 - \frac{\sigma_t^2}{pq} \right)^{1/t} \right]}$ with $t=1$ and $p=0.5$ to yield $N_{ev} = 2.083$ (3 points)

c. (7 points) What is the variance effective size in the total population consisting of all 10 demes?

First use $f_{st} = \sigma_p^2 / (\bar{p}\bar{q})$ to calculate $f_{st} = 0.06/(0.25) = 0.24$ (3 points)

Use $N_{evT} = \frac{nN}{1 - f_{st}}$, so $N_{evT} = 10(2.083)/(1-0.24) = 27.41$ (4 points)

d. (5 points) Suppose that random mating now occurs within each of the 10 isolated demes, and each deme produces an effectively infinite number of offspring. What is the expected deviation from Hardy-Weinberg genotype frequencies (measured by f) in the total population (the offspring of all 10 demes pooled together)?

This is the Wahlund effect, so $f = f_{st} = 0.24$ because $f_{is} = 0$ (random mating within demes)

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5. A neutral, autosomal microsatellite locus has a mutation rate of 10^{-3} per generation.

a. (6 points) In a population of inbreeding effective size 100 and variance effective size of 150, calculate the probability of coalescence before mutation given that either mutation or coalescence occurs for two randomly drawn copies of this locus.

Prob.(mutation before coalescence/mutation or coalescence)

Use
$$= \frac{2xN_{ef}\mu - 2\mu}{2xN_{ef}\mu - 3\mu + 1} \approx \frac{2xN_{ef}\mu}{2xN_{ef}\mu + 1} = \frac{\theta}{\theta + 1}$$
 First, $\theta = 2(2)(100)(10^{-3}) = 0.4$ (3 points)

then, Prob.(coal. before mutation/coal. or mutation) = $1 - \theta / (\theta + 1) = 1 - 0.4 / 1.4 = 0.7143$ (3 points)

b. (3 points) Same problem as a), but now assume that the variance effective size is 50.

Nothing is changed as the calculations depend only on the inbreeding effective size.

c. (2 points) What is the rate of neutral evolution at this locus?

Use Rate = $\frac{1}{2N} \times 2N\mu = \mu = 10^{-3}$.

6. A DNA region of 9 nucleotides is sequenced, with variable sites being found that define 5 haplotypes as follows:

1. A C C G T T G C A
2. A C G G T C G C A
3. A C G G T T G C A
4. A C C G T T A C A
5. A C G G T C G T C

a. (3 points) Construct a cladogram of the haplotypes assuming no recombination and using statistical parsimony.

4-1-3-2-0-5

Suppose that sequence 5 came from another species and can be regarded as a good “outgroup” for the remaining “ingroup” haplotypes.

b. (4 points) Under an isolation by distance model, which haplotype in the ingroup species would be expected to have the most restricted geographical distribution and which the broadest?

Since 5 is the outgroup, haplotype 2 is the oldest and 4 the youngest in the ingroup. Under isolation by distance, we expect the geographical range of a haplotype to be positively correlated with its age, so haplotype 4 should have the most restricted geographical range and haplotype 2 the broadest.

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7. (25 points) Describe the impact and implications of genotype-by-environment and gene-by-gene interactions on the relationship between genotype and phenotype, and give actual (not hypothetical) examples of each type of interaction. Explain the importance of such interactions in making the distinction between cause-and-effect versus cause of variation (use the other side of this page if necessary).