Activity 10-Effects of recessive versus dominant alleles

This activity uses the population genetic simulation program PopGen to explore how being dominant or recessive affects the evolutionary dynamics of an allele under selection.

To receive credit for this activity, do all of the following by Tuesday, Feb. 24 at 9:15 am:
1) Run the simulations specified below and answer the questions after each.
2) Report simulation results in online form (instructions at end)
3) Bring your graphs to class

Go to the following website:
http://www.radford.edu/~rsheehy/Gen_flash/popgen/
It should look familiar from our study of genetic drift.

General instructions for using the simulation program:
The top bar (in blue) contains boxes where you will enter values for the population. To run simulations, use the red GO button on the bottom left. To run more generations, hit CONTINUE

Step 1: setting parameters
To explore the effects of genetic drift alone, you will be testing the effects of parameters on the left side of the blue bar. Hold the following values constant in all simulations:
- Finite Pop. unchecked
- Migration unchecked
- Mutation rates 0
- Bottle Neck? unchecked
- # of Populations 5

Fitness: These values refer to the relative fitnesses of different genotypes. By convention, the genotype with the highest probability of surviving (and reproducing) is given a fitness of 1. The relative fitness then indicates how much less likely an individual of another genotype is to survive and reproduce relative to the most fit genotype in the population.

REMEMBER TO HIT THE RESET BUTTON BETWEEN RUNS!

In these simulations, also pay attention to the thin black line shown on the graphs; that line shows what would happen without any effect of genetic drift.

Dominance occurs when the heterozygote has the same phenotype (and therefore the same fitness) as one of the homozygotes. A common misconception about dominance and evolution is that dominant alleles are more common or that they are more likely to be fixed. In
the simulations we did in Activity 8, neither allele was dominant or recessive, because the heterozygote always had a different fitness than either of the homozygotes. In this exercise, we’ll look at the effects of dominance vs. recessiveness on the evolutionary dynamics of a population.

**Condition 1: Recessive allele is deleterious**

Why do deleterious recessive alleles (including lethal alleles) turn out to be relatively common in natural populations? In Activity 8, you generally observed that alleles that decreased fitness were rapidly lost from the population. Use the following simulation and questions to figure out why recessive alleles persist in populations.

Set the following parameters:
- fitness of A1A1 = 1
- fitness of A1A2 = 1
- fitness of A2A2 = 0
- initial frequency A1 = 0.5
- population size: 1000

**(1a)** What does a relative fitness of 0 for the A2A2 genotype mean?

**(1b)** Run the simulation until fixation has occurred in all 5 populations (or, you can give up after 1000 generations, if that hasn’t happened). On average, how many generations does it take to fix the A1 allele? Record your results in the table below and graph the curve on graph 1 (on the last page). Label the line! (Be sure to add a scale to the x-axis, too.)

**(1c)** For a population of 10,000 individuals, how many homozygous recessive individuals do you expect to be born in an average population when the frequency of the A1 allele has risen to 0.99? Use this to explain why loss of a deleterious recessive allele is slow.

**(1d)** Change the fitness of A1A2 to 0.99. What does this fitness value mean? What is the time to fixation now? Record the results in the table below and add them to Graph 1. Label the line!

**(1e)** Why is selection so much more effective, even though the fitness difference between the A1A2 phenotypes in the two simulations is very small?

**(1f)** The first simulations focused on dynamics once an allele was already common; what about when an advantageous allele introduced into the population is dominant? Change the A1A2 fitness back to 1; you should have the following fitnesses: A1A1=1, A1A2 = 1, A2A2=0. From an initial frequency of 0.01, how many generations does it take to reach a frequency of 0.5? (Hint: you may need to decrease the number of generations to see this.) Record that number here: 

**(1g)** Explain why the number in 1f is so low.

**(1h)** Compare the results for fitness A2A2 = 0.9 instead of 0. How does this change the dynamics at low frequencies of the A1 allele? What about at higher frequencies? Add your results to the table and graph 1 and label the lines.
Condition 2: Recessive allele is beneficial
How does being recessive affect beneficial alleles?

Set the following parameters
- fitness of A1A1 = 1
- fitness of A1A2 = 0.9
- fitness of A2A2 = 0.9
- initial frequency A1 = 0.5
- population size: 1000

(2a) Explain why the A1 allele is recessive, given these fitness values.

(2b) Run the simulation until the A1 allele is fixed. How rapidly does this occur? Record the results of this simulation in the table below and graph them on graph 2. Label the line on your graph.

(2c) Now, experiment with the initial frequency to find the lowest initial A1 allele frequency which has about a 50% chance of fixation. To graph 2, add (and label!) the black line that traces the expected result without drift (i.e., the population called infinity in the legend).

Record the initial A1 allele frequency here _________________

(2d) At the allele frequency you identified, how many homozygous recessive individuals are expected to occur in the population?

Record the expected number of homozygous recessive individuals ________________

(2e) How does the relative fitness of the dominant phenotype affect the fate of beneficial recessive alleles? Change the fitness of the dominant phenotype to 0.1. (Leave the initial A1 allele frequency at the value found in 2c.) Record your results in the table below and on graph 2. Label the line on your graph. Compare to the graphs from 2b and 2c and explain the differences.

Overall comparison of the effects of being dominant or recessive:
Compare the graphs from parts 1 and 2. In what ways are they similar? In what ways are they different? What does this show about the effects of whether a beneficial allele is dominant or recessive on its evolutionary dynamics?

<table>
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<tr>
<th>Simulation</th>
<th>initial A1 frequency</th>
<th>fitness A1A1</th>
<th>fitness A1A2</th>
<th>fitness A2A2</th>
<th># fixed</th>
<th>Mean generations to fixation</th>
<th># lost</th>
<th>Mean generations to loss</th>
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<tbody>
<tr>
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Use this link [http://goo.gl/lkrD5o](http://goo.gl/lkrD5o) to go to the online reporting form for your results. For credit, this must be completed by 9:15 am on Tuesday, Feb. 24.

Remember to bring your graphs to class!
Graph 1: Selection on deleterious recessive alleles

A1 allele frequency

Generations

Graph 2: Selection on beneficial recessive alleles

A1 allele frequency

Generations

0%  25%  50%  75%  100%

0%  25%  50%  75%  100%

2245 W? (check if yes)

_____________________ Last Name

_____________________ SID