

# An introduction to population genetics

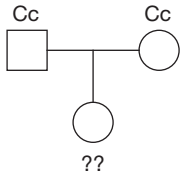
Population genetics is the result of a marriage of Mendelian genetics with Darwinian natural selection. Mendelian genetics allows us to predict the characteristics of the next generation within families. Population genetics will allow us to predict the characteristics of the next generation in whole populations.

## Genes in populations

Recall that in Mendelian genetics, we predict the genotypes of offspring by tracking alleles from parents to offspring. If both parents are carriers

## 2


## An introduction to population genetics



Possible eggs

	C	c
Possible sperm	C	Cc
	c	cc

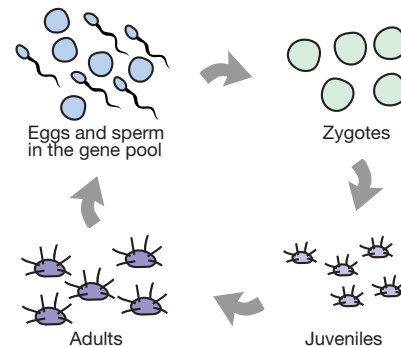
Zygotes



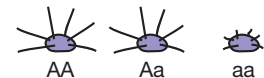
A small diagram of a sperm cell with a head and a tail.

for cystic fibrosis, for example, they have genotype Cc. We can use a Punnett square to predict that they will have offspring with genotypes CC, Cc, and cc in a 1:2:1 ratio, and that the probability that any particular child will have cystic fibrosis is 1 in 4.

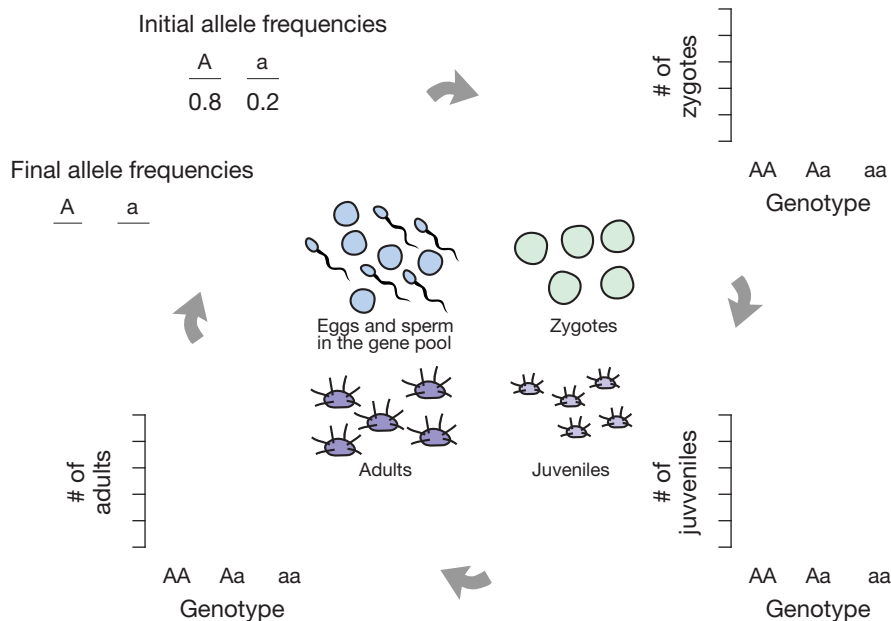
In population genetics, we will track alleles and genotypes across the entire life cycle of a population. Sea urchins are a useful organism to think about. Sea urchins don't copulate. Instead, males and females simply release their sperm and eggs to float on the tide. The eggs and sperm, which we will think of as the **gene pool**, find each other more or less at random and join to become zygotes. We will follow the zygotes as they grow into juveniles, then adults. Finally, we will watch the adults release eggs and sperm to produce the next generation's gene pool:



Imagine a population of sea urchins in which a trait, like spine length, is controlled by two alleles of a single gene. The relationship between genotype and phenotype might work as shown at right.



Imagine that in the urchin population's gene pool, 80% of the eggs and sperm carry allele *A*, and 20% carry allele *a*. The **frequency** of *A* is 0.8, and the frequency of *a* is 0.2. If the eggs and sperm combine at random, what will be the frequencies of *AA*, *Aa*, and *aa* among the zygotes? The juveniles? The adults? What will be the frequency of alleles *A* and *a* in the next generation's gene pool?



One way to answer these questions is by simulation. Open PopCycle. The ad for Evolutionary Analysis will go away after a few seconds; you can dismiss it sooner by clicking on it. You should see a gene pool

in the upper left corner of the window, with eggs and sperm floating and swimming around in it. The darker gametes carry allele  $A$ ; the lighter ones carry allele  $a$ .

The allele frequencies in the default gene pool are 0.5 for  $A$  and 0.5 for  $a$ . We want to simulate a population in which the frequencies are 0.8 and 0.2. Enter 0.8 as the new value for the frequency of  $A$  and hit the return key. You should see the gene pool change.

Now to make zygotes. If you click anywhere in the gene pool, Pop-Cycle will pick an egg from the gene pool at random. Click again and it will pick a sperm. You will see the zygote's genotype displayed briefly on the screen. Then the zygote you've just made is added to the bar graph at upper right and the egg, sperm, and zygote genotype displays clear themselves. Now do the following:

1. Keep clicking on the gene pool until you have made 100 zygotes. Then click on the Done button. The bars on the graph should turn green. Record the number of zygotes of each genotype:

AA:            Aa:            aa:

2. We are going to imagine that all of the zygotes survive to become juveniles. Make sure that the survival rates are set to 1.0 for all three genotypes. Then click on the Done button. There should be no surprises. Record the number of juveniles of each genotype:

AA:            Aa:            aa:

3. We are also going to imagine that the juveniles become adults without any urchins moving into or out of the population. Make sure the number of migrants is set to zero for all three genotypes. Then click on the done button. Again, no surprises. Record the number of adults of each genotype:

AA:            Aa:            aa:

4. Finally, we are going to imagine that when the adults make gametes for the new gene pool, they do so without any mutations that might turn copies of allele *A* into *a* or vice versa. Make sure that the mutation rates are both set to zero—then wait.

Before you click the Done button, predict what the new allele frequencies are going to be. Imagine that each of your adults makes 10 gametes for the new gene pool. Fill in the table below:

The \_\_\_\_ *AA* adults make a total of \_\_\_\_ *A* gametes and \_\_\_\_ *a* gametes.

The \_\_\_\_ *Aa* adults make a total of \_\_\_\_ *A* gametes and \_\_\_\_ *a* gametes.

The \_\_\_\_ *aa* adults make a total of \_\_\_\_ *A* gametes and \_\_\_\_ *a* gametes.

What is the total number of gametes in the new gene pool? \_\_\_\_

What is the total number of gametes carrying allele *A*? \_\_\_\_

What is the frequency of allele *A* in the new gene pool? \_\_\_\_

## An introduction to population genetics

Now click on the Done button to see the frequency of  $A$  in the new gene pool. Were you right? If not, try to figure out why.


- Recall that in our original gene pool the frequency of allele  $A$  was 0.8. Has it changed in the new gene pool? If so, why?
- Should we have expected the frequency of allele  $A$  to change from one generation to the next? Think back to our starting gene pool, with a frequency of 0.8 for allele  $A$  and 0.2 for allele  $a$ . If we make 100 zygotes by drawing gametes at random, how many zygotes of each genotype should we expect? We can answer this question the same way we predicted the genotype frequencies for cystic fibrosis within a single family: with a Punnett Square. We just have to use a bigger one. We put eggs along one side in a ratio of 8 to 2, and sperm along the other side in a ratio of 8 to 2. Then we fill in the zygotes. Fill in the zygotes in the Punnett Square below:

○ Possible eggs

	A	A	A	A	A	A	A	A	a	a
A										
A										
A										
A										
A										
A										
A										
a										
a										

Zygotes

Possible sperm



How many zygotes of each genotype do you expect?:

AA:

Aa:

aa:

You can check your answer by going back to PopCycle. Reset the simulation by selecting Reset under the File menu. Change the frequency of allele *A* to 0.8. Now click on the Ideal button. The zygote bar graph now shows you how many zygotes of each genotype we should expect. If your Punnett square prediction was not correct, try to figure out why.

7. Shepherd the zygotes from the ideal population around their life cycle—to juveniles with 100 % survival, to adulthood with no migration, and to the new gene pool with no mutation. Has the frequency of allele *A* changed from its starting value of 0.8? Why or why not?
8. Think a bit more about why your first simulation might have produced genotype frequencies among the zygotes that were different from our ideal expectations. Reset PopCycle and once again set the frequency of allele *A* in the gene pool to 0.8. Now start making zygotes by clicking on the gene pool. Watch the graph of zygote genotypes as it grows. As you make more and more zygotes, how do the genotype frequencies compare to the proportions of the ideal expectation?

## Genes in populations with selection

What happens when we add natural selection to our simulation?

9. Reset PopCycle, set the frequency of allele  $A$  to 0.8, and click on the Ideal button. Now, before clicking done to make the zygotes grow into juveniles, change the survival rates. Leave the rate for  $AA$  at 1.0, but set the rate for  $Aa$  to 0.5, and the rate for  $aa$  to 0.0. As you change the rates, you will see the juvenile graph change to show what is going to happen. When you have the rates set, click Done. Record the number of juveniles of each genotype:

AA:                      Aa:                      aa:

10. Make sure the number of migrants is zero for all genotypes, then click Done. Make sure the mutation rates are set to zero, but before you click Done, predict what's going to happen:

The \_\_\_\_  $AA$  adults make a total of \_\_\_\_  $A$  gametes and \_\_\_\_  $a$  gametes.

The \_\_\_\_  $Aa$  adults make a total of \_\_\_\_  $A$  gametes and \_\_\_\_  $a$  gametes.

The \_\_\_\_  $aa$  adults make a total of \_\_\_\_  $A$  gametes and \_\_\_\_  $a$  gametes.

What is the total number of gametes in the new gene pool? \_\_\_\_

What is the total number of gametes carrying allele  $A$ ? \_\_\_\_

What is the frequency of allele  $A$  in the new gene pool? \_\_\_\_



Now click on the Done button to see the frequency of  $A$  in the new gene pool. Were you right? Has the frequency of allele  $A$  changed from its starting value of 0.8? Why or why not?

## Summary

We can now define **evolution** as change across generations in the frequencies of alleles. And we can explain it with Darwin's theory of evolution by natural selection, phrased in the jargon of Mendelian genetics:

1. Individuals in a population vary in **phenotype** (= differ in form, physiology, etc.).
  2. Some of the variation is **heritable** (= differences in phenotype are at least partly due to differences in **genotype**). The ultimate source of genetic variation is mutation.
  3. More offspring are born than will survive to reproduce
  4. Individuals with some genotypes survive and/or reproduce at higher rates than do individuals with other genotypes (= natural selection)
- Conclusion: Allele frequencies change across generations.

What else, besides differences in survival, can make the frequency of allele  $A$  change? Play with PopCycle. Can migration do it? How easily? How about mutation?