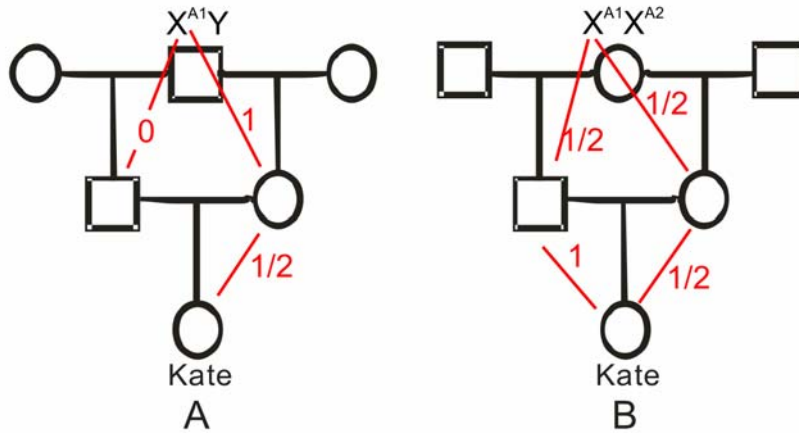


1. Consider the following two pedigrees:



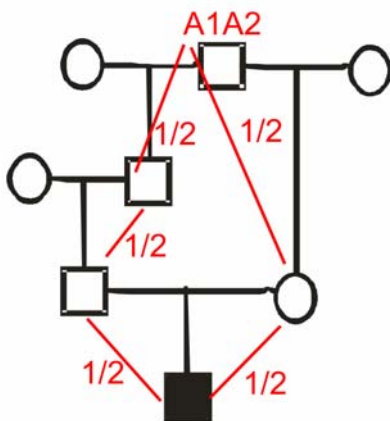
Ignore all ancestors that are not shared by Kate's parents

What are the probabilities of identity by descent in individual "Kate" for a randomly chosen X-linked allele in pedigrees A and B?

A:  $F=0$ , because Kate's father gets the Y chromosome from his father and not  $X^{A1}$ .  
 Kate cannot be  $X^{A1} X^{A1}$  homozygous.

B:  
 Probability that Kate is  $X^{A1} X^{A1}$  homozygous is  $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$   
 Probability that Kate is  $X^{A2} X^{A2}$  homozygous is  $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{8}$   
 $F = P(X^{A1} X^{A1}) + P(X^{A2} X^{A2}) = \frac{1}{8} + \frac{1}{8} = \frac{1}{4}$

2. Consider the pedigree below:



What is the pedigree inbreeding coefficient  $F$  of the male individual shown as a black square? Assume that all relevant genetic relationships are shown in the pedigree.

Probability that Johnny is  $A1A1$  homozygous is  $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{32}$   
 Probability that Johnny is  $A2A2$  homozygous is  $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{32}$

$F = P(A1A1) + P(A2A2) = \frac{1}{16}$

3. Consider a fictional scenario:

The dominant **allele L** codes for lactase persistence. People who are recessive homozygotes (ll) cannot digest milk as adults; dominant homozygotes (LL) and heterozygotes (Ll) have no problem digesting milk. Among **200** people from Eastern Mongolia, **32** were unable to digest milk.

a. Assuming that the population does not deviate from a **Hardy-Weinberg equilibrium**, estimate the frequencies of the alleles **L** and **l**:

$$\text{fr}(ll) = 32/200 = 0.16 = q^2$$

$$\text{fr}(l)_0 = \sqrt{q^2} = \sqrt{0.16} = 0.4 \qquad \text{fr}(L)_0 = p = 1 - q = 0.6$$

b. Milk in Mongolia is a common source of bacterial infection. Individuals who drink milk are very likely to contract a severe stomach infection and die. The mortality rate from stomach infection among milk drinkers (the LL and Ll genotypes) is 40%.

Using this information and the allele frequencies you have found above, fill in the table : (2 pt)

	LL	Ll	ll	total
frequency of each genotype	$p^2=0.36$	$2pq=0.48$	$q^2=0.16$	1
relative fitness ( $w$ )	0.6	0.6	1	
coefficient of selection ( $s$ )	0.4	0.4	1	
genotype frequencies after selection	$\frac{p^2 \times 0.6}{0.6p^2 + 0.6 \times 2pq + q^2} = 0.22/0.66 = 0.34$	$\frac{2pq \times 0.6}{0.6p^2 + 0.6 \times 2pq + q^2} = 0.29/0.66 = 0.44$	$0.16/0.66 = 0.24$	1

c. Find the respective allele frequencies after selection:

$$\text{fr}(l)_1 = \text{fr}(ll) + 0.5\text{fr}(Ll) = 0.24 + 0.22 = 0.46 \qquad \text{fr}(L)_1 = 1 - \text{fr}(l) = 0.54$$

d. What type of natural selection is operating on this population?: \_\_\_\_\_

**Directional selection for the recessive phenotype**

f. What are the points of equilibrium for this type of natural selection? Which are stable and which are unstable? Why?

$$\Delta q = q^2ps / (1 - p^2s - 2pqs); \Delta q = 0 \text{ when } q=0 \text{ or } p=0;$$

when  $0 < q < 1$ ,  $\Delta q > 0 \rightarrow q$  is increasing  $\rightarrow \hat{q} = 0$  is unstable,  $\hat{q} = 1$  is stable

4. The recessive allele *c* codes for cystic fibrosis. People who are recessive homozygotes (*cc*) have a tendency toward chronic lung infections. Mortality among people with cystic fibrosis is 15% greater than that of people without the disease. Dominant homozygotes show no symptoms of the disease. In a randomly mating population, 9% of newborns were diagnosed with cystic fibrosis.

A. Estimate the frequency of allele *c*, assuming a Hardy-Weinberg equilibrium:

We assume that the population of newborns does not deviate from the Hardy-Weinberg

expectations: Thus,  $fr(c)_0 = q = \sqrt{q^2} = \sqrt{0.09} = 0.3$   $fr(C) = 1 - 0.3 = 0.7$

B. What type of natural selection is operating?: **Directional selection for the dominant phenotype**

C. Calculate allele frequencies in this case after one and after two generations:

$$\Delta q = q_1 - q \rightarrow q_1 = q + \Delta q$$

In the case of directional selection for the dominant phenotype

$$\Delta q = -q^2ps / (1 - q^2s) \quad [q^2 = 0.09, p = 0.7, s = 0.15]$$

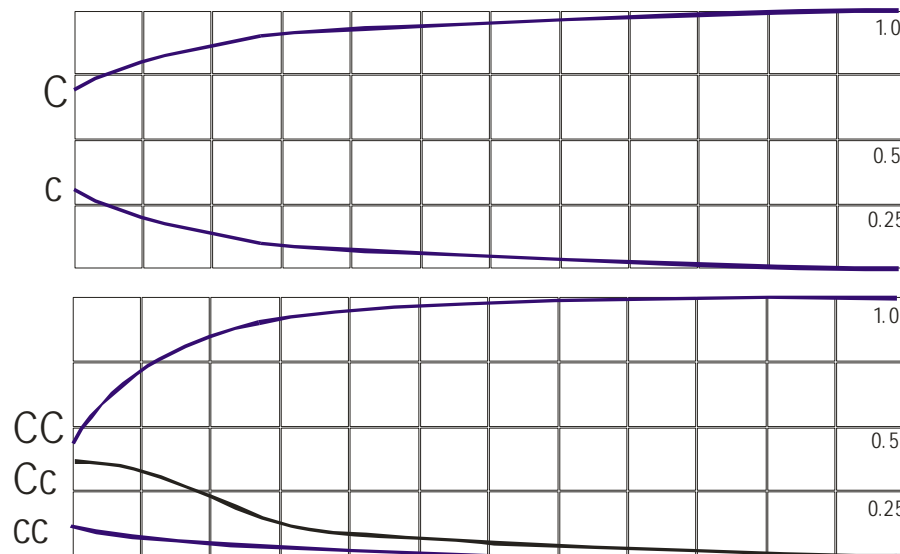
$$\Delta q = -0.09 * 0.7 * 0.15 / (1 - 0.09 * 0.15) = -0.0096$$

$$q_1 = q + \Delta q = 0.3 - 0.0096 = \mathbf{0.29}$$

$$q_2 = q_1 + \Delta q_1 = 0.29 - [0.29^2 * 0.71 * 0.15 / (1 - 0.29^2 * 0.15)] = 0.29 - 0.009 = \mathbf{0.28}$$

D. Predict how the allele and genotype frequencies in this case are likely to change through time.

Make rough graphs:



5. For a certain type of balancing selection it was found that:

$$\Delta q = pq(0.6p - 0.4q)/(1 - 0.6p^2 - 0.4q^2)$$

Find three points of equilibrium:

Which of them are stable and which of them are unstable?

For balancing selection,  $\Delta q = pq(s_1p - s_2q)/(1 - p^2s_1 - q^2s_2) \rightarrow s_1=0.6, s_2=0.4 \rightarrow$

$$\hat{q}_{st} = s_1/(s_1 + s_2) = 0.6/(0.6+0.4) = 0.6$$

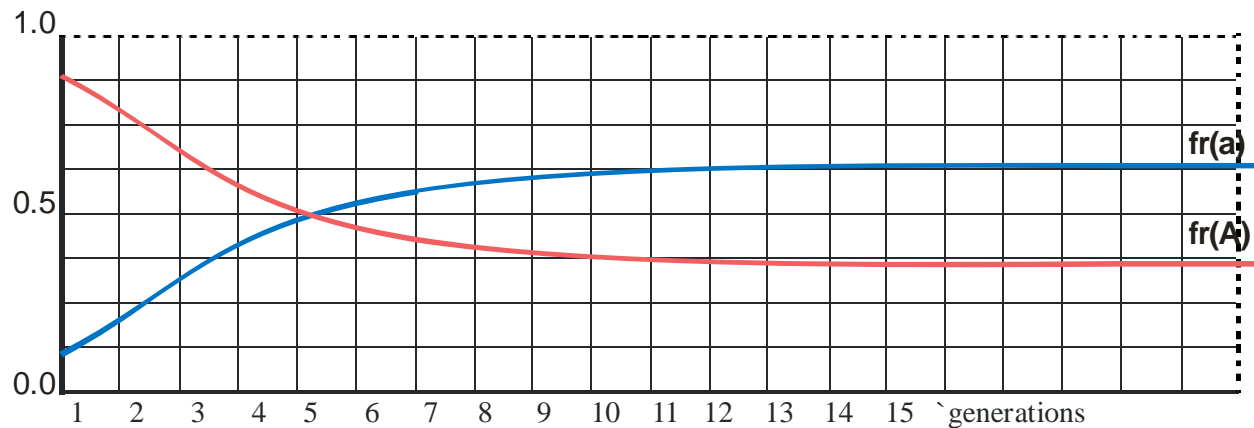
$\hat{q} = 0$  and  $1$  are unstable

What are the genotype frequencies at equilibrium?

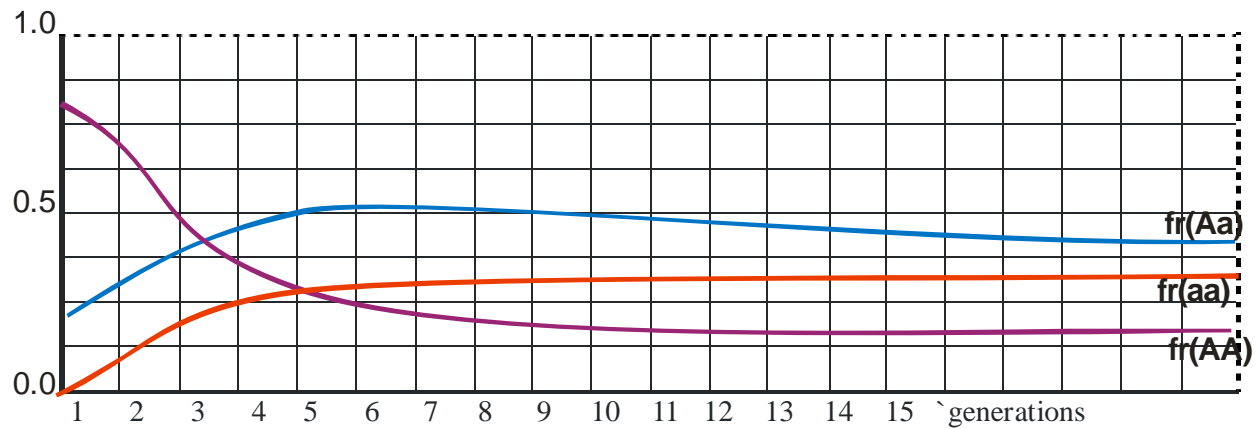
$$fr(AA) = \underline{0.16} \quad fr(Aa) = \underline{0.48} \quad fr(aa) = \underline{0.36}$$

In the population of western Colorado, the frequency of allele **a** is 0.1.

Make an approximate chart showing how allele frequencies will change over the generations:



Make an approximate chart of the change in genotype frequencies over the generations:



6. Consider another fictional scenario:

Among a group of hunting and gathering native Australians, individuals who can detect a bitter taste in certain plants, even when the pertinent alkaloids are present in very low concentrations (**TT** homozygotes), are highly valued as plant gatherers. Non-tasters (**tt** homozygotes) don't even try to gather plants and pursue hunting instead. **Tt** heterozygotes try to gather plants, but often die from ingesting toxins. This situation results in the following values of relative fitness:

	<b>TT</b>	<b>Tt</b>	<b>tt</b>
<b>W</b>	1	0.2	0.8

- What type of natural selection is operating? **Disruptive**
- Find all points of equilibrium. Which of them are stable?

$$s_1 = 1 - w_1 = 1 - 0.2 = 0.8, \quad s_2 = 1 - w_2 = 1 - 0.8 = 0.2$$

$$\Delta p = \frac{p^2 + (1 - s_1)pq}{1 - 2pqs_1 - q^2s_2} - p = \frac{p^2 + (1 - s_1)pq - p + 2p^2qs_1 + q^2ps_2}{1 - 2pqs_1 - q^2s_2}$$

$$\Delta p = 0, \text{ when, } p^2 + (1 - s_1)pq - p + 2p^2qs_1 + q^2ps_2 = 0$$

This simplifies:

$$pq(s_1 - 2s_1q + s_2q) = 0 \Rightarrow \Delta p = 0, \text{ when}$$

$$p = 0, q = 0, \text{ or } s_1 - 2s_1q + s_2q = 0 \Rightarrow q = \frac{s_1}{2s_1 - s_2}$$

$$\text{Three points of equilibrium: } \hat{q} = 1, \hat{q} = 0, \hat{q} = \frac{0.8}{2 \times 0.8 - 0.2} = 0.57$$

When  $0 < q < 0.57$ ,  $\Delta q < 0 \rightarrow q$  is declining

when  $0.57 < q < 1$ ,  $\Delta q > 0 \rightarrow q$  is growing  $\rightarrow \hat{q} = 1$  is stable,  $\hat{q} = 0$  is stable,  $\hat{q} = 0.57$  is unstable

- The current frequency of allele **t** is 0.55. Make an approximate chart of allele frequency change in the tribe over generations.

$fr(t) < 0.57 \rightarrow \Delta q < 0 \rightarrow fr(t)$  is going to decline

