Gene Pool Exercise

Cut out each of the beetles on this page and use them to reenact different events within a gene pool as described in this topic (see Gene Pools and Evolution, Changes in a Gene Pool, Founder Effect, Population Bottlenecks, Genetic Drift).
Gene Pools and Evolution

The diagram below illustrates the dynamic nature of gene pools. It portrays two imaginary populations of one beetle species. Each beetle is a 'carrier' of genetic information, represented here by the alleles (A and a) for a single codominant gene that controls the beetle's color. Normally, there are three versions of the phenotype: black, dark, and pale. Mutations may create other versions of the phenotype. Some of the microevolutionary processes that can affect the genetic composition (allele frequencies) of the gene pool are illustrated below.

Immigration: Populations can gain alleles when they are introduced from other gene pools. Immigration is one aspect of gene flow.

Natural selection: Selection pressure against certain allele combinations may reduce reproductive success or lead to death. Natural selection sorts genetic variability and accumulates and maintains favorable genotypes in a population. It tends to reduce genetic diversity within the gene pool and increase differences between populations.

Gene flow: Genes are exchanged with other gene pools as individuals move between them. Gene flow is a source of new genetic variation and tends to reduce differences between populations that have accumulated because of natural selection or genetic drift.

Mutations: Spontaneous mutations can develop that alter the allele frequencies of the gene pool, and even create new alleles. Mutation is very important to evolution, because it is the original source of genetic variation that provides new material for natural selection.

Emigration: Genes may be lost to other gene pools.

The term deme describes a local population that is genetically isolated from other populations in the species. Demes usually have some clearly definable genetic or other character that sets them apart from other populations.

Geographical barriers: Isolate the gene pool and prevent regular gene flow between populations.

Key to genotypes and phenotypes
- AA: Black Homozygous dominant
- Aa: Dark Heterozygous
- aa: Pale Homozygous recessive
- A'A': Mottled Homozygous dominant (mutant)

Gene drift: Chance events can cause the allele frequencies of small populations to "drift" (change) randomly from generation to generation. Genetic drift can play a significant role in the microevolution of very small populations. The two situations most often leading to populations small enough for genetic drift to be significant are the bottleneck effect (where the population size is dramatically reduced by a catastrophic event) and the founder effect (where a small number of individuals colonize a new area).

Mate selection (non-random mating): Individuals may not select their mates randomly and may seek out particular phenotypes, increasing the frequency of these "favored" alleles in the population.
Factors Affecting Gene Pools

One of the fundamental concepts for population genetics is stated as follows:

For a very large, randomly mating population, the proportion of dominant to recessive alleles remains constant from one generation to the next (the population is in genetic equilibrium).

In practical terms this means that, if a gene pool is to remain unchanged, it must satisfy all of the criteria listed on the left side of the diagram below (factors that favor gene pool stability). The fact that few populations can be identified as meeting all (or any) of these criteria means that they must be undergoing continual change in their genetic makeup.

For each of the five factors (numbers 1-5) below, state briefly how and why each would affect the allele frequency in a gene pool:

1. Population size:

2. Mate selection:

3. Gene flow between populations:

4. Mutations:

5. Natural selection:

6. List the factors that tend to:
   (a) Increase genetic variation in populations:
   (b) Decrease genetic variation in populations:
Changes in a Gene Pool

The diagram below shows an imaginary population of beetles undergoing changes as it is subjected to two events. The three phases represent a progression in time, i.e., the same gene pool, undergoing change. The beetles have three phenotypes determined by the amount of pigment deposited in the cuticle. Three versions of this trait exist: black, dark, and pale. The gene controlling this character is represented by two alleles, A and a. Your task is to analyze the gene pool as it undergoes changes.

Phase 1: Initial gene pool

Calculate the frequencies of the allele types and allele combinations by counting the actual numbers, then working them out as percentages.

<table>
<thead>
<tr>
<th>Allele types</th>
<th>Allele combinations</th>
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<tbody>
<tr>
<td>A</td>
<td>aa</td>
</tr>
<tr>
<td>a</td>
<td>AA, Aa</td>
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<tr>
<td>AA</td>
<td>aa</td>
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<tr>
<td>Aa</td>
<td>AA, Aa</td>
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No.

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<tr>
<th></th>
<th>A</th>
<th>a</th>
<th>AA</th>
<th>Aa</th>
<th>aa</th>
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<tbody>
<tr>
<td></td>
<td>27</td>
<td>7</td>
<td>54</td>
<td>28</td>
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Phase 2: Natural selection

In the same gene pool at a later time, there was a change in the allele frequencies. This was due to the loss of certain allele combinations due to natural selection. Some of those with a genotype of aa were eliminated (poor fitness).

Calculate as for above. Do not include the individuals surrounded by a small white arrow; they are dead!

<table>
<thead>
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<tbody>
<tr>
<td>A</td>
<td>aa</td>
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<td>a</td>
<td>AA, Aa</td>
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<tr>
<th></th>
<th>A</th>
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%

Phase 3: Immigration and emigration

This particular kind of beetle exhibits wandering behavior. The allele frequencies change again due to the introduction and departure of individual beetles, each carrying certain allele combinations.

Calculate as above. In your calculations, include the individual coming into the gene pool (AA), but remove the one leaving (aa).

<table>
<thead>
<tr>
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</tr>
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<tbody>
<tr>
<td>A</td>
<td>aa</td>
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<td>a</td>
<td>AA, Aa</td>
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No.

<table>
<thead>
<tr>
<th></th>
<th>A</th>
<th>a</th>
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%

1. Explain how the number of dominant alleles (A) in the genotype of a beetle affects its phenotype:

2. For each phase in the gene pool above (place your answers in the tables provided; some have been done for you):

   (a) Determine the relative frequencies of the two alleles: A and a. Simply total the A alleles and a alleles separately.
   (b) Determine the frequency of how the alleles come together as allele pair combinations in the gene pool (AA, Aa, and aa). Count the number of each type of combination.
   (c) For each of the above, work out the frequencies as percentages:

\[
\text{Allele frequency} = \frac{\text{Number of counted alleles}}{\text{Total number of alleles}} \times 100
\]
The Hardy-Weinberg equation provides a simple mathematical model of genetic equilibrium in a gene pool, but its main application in population genetics is in calculating allele and genotype frequencies in populations, particularly as a means of studying changes and measuring their rate. The use of the Hardy-Weinberg equation is described below.

\[(p + q)^2 = p^2 + 2pq + q^2 = 1\]

**Punnett square**

- Frequency of allele combination AA in the population is represented as \(p^2\)
- Frequency of allele combination aa in the population is represented as \(q^2\)
- Frequency of allele combination Aa in the population (add these together to get \(2pq\))

**Frequency of allele types**
- \(p = \text{Frequency of allele } A\)
- \(q = \text{Frequency of allele } a\)

**Frequency of allele combinations**
- \(p^2 = \text{Frequency of } AA \text{ (homozygous dominant)}\)
- \(2pq = \text{Frequency of } Aa \text{ (heterozygous)}\)
- \(q^2 = \text{Frequency of } aa \text{ (homozygous recessive)}\)

The Hardy-Weinberg equation is applied to populations with a simple genetic situation: dominant and recessive alleles controlling a single trait. The frequency of all of the dominant (\(A\)) and recessive alleles (\(a\)) equals the total genetic complement, and adds up to 1 or 100% of the alleles present.

**How To Solve Hardy-Weinberg Problems**

In most populations, the frequency of two alleles of interest is calculated from the proportion of homozygous recessives (\(q^2\)), as this is the only genotype identifiable directly from its phenotype. If only the dominant phenotype is known, \(q^2\) may be calculated \((1 - \text{the frequency of the dominant phenotype})\). The following steps outline the procedure for solving a Hardy-Weinberg problem:

**Remember that all calculations must be carried out using proportions, NOT PERCENTAGES!**

1. Examine the question to determine what piece of information you have been given about the population. In most cases, this is the percentage or frequency of the homozygous recessive phenotype \(q^2\), or the dominant phenotype \(p^2 + 2pq\) (see note above).

2. The first objective is to find out the value of \(p\) or \(q\), if this is achieved, then every other value in the equation can be determined by simple calculation.

3. Take the square root of \(q^2\) to find \(q\).

4. Determine \(p\) by subtracting \(q\) from 1 (i.e. \(p = 1 - q\)).

5. Determine \(p^2\) by multiplying \(p\) by itself (i.e. \(p^2 = p \times p\)).

6. Determine \(2pq\) by multiplying \(p\) times \(q\) times 2.

7. Check that your calculations are correct by adding up the values for \(p^2 + q^2 + 2pq\) (the sum should equal 1 or 100%).

**Worked example**

In the American white population approximately 70% of people can taste the chemical phenylthiocarbamide (PTC) (the dominant phenotype), while 30% are non-tasters (the recessive phenotype).

**Determine the frequency of:**

(a) Homozygous recessive phenotype (\(q^2\)), 30% – provided.

(b) The dominant allele (\(p\)).

(c) Homozygous tasters (\(p^2\)).

(d) Heterozygous tasters (\(2pq\)).

**Data:** The frequency of the dominant phenotype (70% tasters) and recessive phenotype (30% non-tasters) are provided.

**Working:**

- Recessive phenotype: \(q^2 = 0.30\) - use 0.30 for \(q\) calculation.
  - \(\therefore q = 0.5477\) square root of 0.30
  - \(\therefore p = 0.4523\)
  - \(1 - q = p\)
  - \(1 - 0.5477 = 0.4523\)
- Use \(p\) and \(q\) in the equation (top) to solve any unknown.
  - Homozygous dominant: \(p^2 = 0.2046\)
  - \(\therefore p \times p = 0.4523 \times 0.4523\)
  - \(2pq = 0.4853\)

1. A population of hamsters has a gene consisting of 90% M alleles (black) and 10% m alleles (gray). Mating is random.

**Data:** Frequency of recessive allele (10% m) and dominant allele (90% M).

Determine the proportion of offspring that will be black and the proportion that will be gray (show your working):

- Recessive allele: \(q = \)
- Dominant allele: \(p = \)
- Recessive phenotype: \(q^2 = \)
- Homozygous dominant: \(p^2 = \)
- Heterozygous: \(2pq = \)
2. You are working with pea plants and found 36 plants out of 400 were dwarf.
   **Data:** Frequency of recessive phenotype (36 out of 400 = 9%)
   (a) Calculate the frequency of the tall gene: ________________
   (b) Determine the number of heterozygous pea plants:

3. In humans, the ability to taste the chemical phenylthiocarbamide (PTC) is inherited as a simple dominant characteristic. Suppose you found out that 360 out of 1000 college students could not taste the chemical.
   **Data:** Frequency of recessive phenotype (360 out of 1000).
   (a) State the frequency of the gene for tasting PTC:
   
   (b) Determine the number of heterozygous students in this population:

4. A type of deformity appears in 4% of a large herd of cattle. Assume the deformity was caused by a recessive gene.
   **Data:** Frequency of recessive phenotype (4% deformity).
   (a) Calculate the percentage of the herd that are carriers of the gene:
   
   (b) Determine the frequency of the dominant gene in this case:

5. Assume you placed 50 pure bred black guinea pigs (dominant allele) with 50 albino guinea pigs (recessive allele) and allowed the population to attain genetic equilibrium (several generations have passed).
   **Data:** Frequency of recessive allele (50%) and dominant allele (50%).
   Determine the proportion (%) of the population that becomes white:

6. It is known that 64% of a large population exhibit the recessive trait of a characteristic controlled by two alleles (one is dominant over the other).
   **Data:** Frequency of recessive phenotype (64%). Determine the following:
   (a) The frequency of the recessive allele:
   
   (b) The percentage that are heterozygous for this trait:
   
   (c) The percentage that exhibit the dominant trait:
   
   (d) The percentage that are homozygous for the dominant trait:
   
   (e) The percentage that has one or more recessive alleles:

7. Albinism is recessive to normal pigmentation in humans. The frequency of the albino allele was 10% in a population.
   **Data:** Frequency of recessive allele (10% albino allele).
   Determine the proportion of people that you would expect to be albino:

   Recessive allele: \( q = 0.1 \)
   Dominant allele: \( p = 0.9 \)
   Recessive phenotype: \( q^2 = 0.01 \)
   Homozygous dominant: \( p^2 = 0.81 \)
   Heterozygous: \( 2pq = 0.18 \)
Occasionally, a small number of individuals from a large population may migrate away, or become isolated from, their original population. If this colonizing or 'founder' population is made up of only a few individuals, it will probably have a non-representative sample of alleles from the parent population's gene pool. As a consequence of this founder effect, the colonizing population may evolve differently from that of the parent population, particularly since the environmental conditions for the isolated population may be different. In some cases, it may be possible for certain alleles to be missing altogether from the individuals in the isolated population. Future generations of this population will not have this allele.

Some individuals from the mainland population are carried at random to the offshore island by natural forces such as strong winds.

This population may not have the same allele frequencies as the mainland population.

Mainland population

<table>
<thead>
<tr>
<th>Allele frequencies</th>
<th>Phenotype frequencies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Actual numbers</td>
<td>Calculate %</td>
</tr>
<tr>
<td></td>
<td>Black</td>
</tr>
<tr>
<td>Allele A</td>
<td></td>
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<tr>
<td>Allele a</td>
<td></td>
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<tr>
<td>Total</td>
<td></td>
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Colonizing Island population

<table>
<thead>
<tr>
<th>Allele frequencies</th>
<th>Phenotype frequencies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Actual numbers</td>
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<tr>
<td>Allele a</td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td></td>
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</tbody>
</table>

1. Compare the mainland population to the population which ended up on the island (use the spaces in the tables above):

(a) Count the phenotype numbers for the two populations (i.e. the number of black, dark and pale beetles).
(b) Count the allele numbers for the two populations: the number of dominant alleles (A) and recessive alleles (a).
   Calculate these as a percentage of the total number of alleles for each population.

2. Describe how the allele frequencies of the two populations are different:

3. Describe some possible ways in which various types of organism can be carried to an offshore island:

(a) Plants:

(b) Land animals:

(c) Non-marine birds:

4. Since founder populations are often very small, describe another process that may further alter the allele frequencies:
Populations may sometimes be reduced to low numbers by predation, disease, or periods of climatic change. A population crash may not be 'selective': it may affect all phenotypes equally. Large scale catastrophic events (e.g. fire or volcanic eruption) are examples of such non-selective events. Humans may severely (and selectively) reduce the numbers of some species through hunting and/or habitat destruction. These populations may recover, having squeezed through a 'bottleneck' of low numbers.

The diagram below illustrates how population numbers may be reduced as a result of a catastrophic event. Following such an event, the small number of individuals contributing to the gene pool may not have a representative sample of the alleles in the pre-catastrophe population, i.e. the allele frequencies in the remnant population may be altered. Genetic drift may cause further changes to allele frequencies. The small population may return to previous levels but with a reduced genetic diversity.

Population Bottleneck in Cheetahs

Until recently, the dwindling population of cheetahs in the wild was thought to be the result of over-hunting and habitat destruction. The world population of cheetahs has declined to fewer than 20,000. Recent genetic analysis has found that the total cheetah population has very little genetic diversity (they all have very similar genotypes). It appears that cheetahs may have narrowly escaped extinction at the end of the last ice age, about 10,000 years ago. The population crash may have been so severe that the total species may have been reduced to a single family group. If all modern cheetahs arose from a single surviving litter, this would explain the lack of genetic diversity. This is not a surprising finding, since 75% of all large mammals perished at this time (including well-known animals such as mammoths, cave bears and sabre-tooth tigers). The lack of genetic variation has led to a number of features that threaten the survival of the cheetah species, including: sperm abnormalities, decreased fecundity (number of offspring produced in its lifetime), high cub mortality, and sensitivity to disease.

1. Endangered species are often subjected to population bottlenecks. Explain how population bottlenecks affect the ability of a population of an endangered species to recover from its plight:

2. Explain why the lack of genetic diversity in cheetahs has increased their sensitivity to disease:

3. Describe the effect of a population bottleneck on the potential of a species to adapt to changes (i.e. its ability to evolve):
Genetic Drift

Not all individuals, for various reasons, will be able to contribute their genes to the next generation. Genetic drift (also known as the Sewell-Wright Effect) refers to the random changes in allele frequency that occur in all populations, but are much more pronounced in small populations. In a small population, the effect of a few individuals not contributing their alleles to the next generation can have a great effect on allele frequencies. Alleles may even become lost from the gene pool altogether (frequency becomes 0%) or fixed as the only allele for the gene present (frequency becomes 100%).

The genetic makeup (allele frequencies) of the population changes randomly over a period of time

<table>
<thead>
<tr>
<th>Generation 1</th>
<th>Generation 2</th>
<th>Generation 3</th>
</tr>
</thead>
<tbody>
<tr>
<td>A = 16 (53%)</td>
<td>A = 15 (50%)</td>
<td>A = 13 (43%)</td>
</tr>
<tr>
<td>a = 14 (47%)</td>
<td>a = 15 (50%)</td>
<td>a = 17 (57%)</td>
</tr>
</tbody>
</table>

This diagram shows the gene pool of a hypothetical small population over three generations. For various reasons, not all individuals contribute alleles to the next generation. With the random loss of the alleles carried by these individuals, the allele frequency changes from one generation to the next. The change in frequency is directionless as there is no selecting force. The allele combinations for each successive generation are determined by how many alleles of each type are passed on from the preceding one.

Computer Simulation of Genetic Drift

Below are displayed the change in allele frequencies in a computer simulation showing random genetic drift. The breeding population progressively gets smaller from left to right. Each simulation was run for 140 generations.

1. Explain what is meant by genetic drift:

2. Describe how genetic drift affects the amount of genetic variation within very small populations:

3. Identify a small breeding population of animals or plants in your country in which genetic drift could be occurring: