20.2: Changes in Allele Frequency

If two individuals mate that are heterozygous (e.g., **Bb**) for a trait, we find that

- 25% of their offspring are homozygous for the dominant allele (**BB**)
- 50% are heterozygous like their parents (**Bb**)
- 25% are homozygous for the recessive allele (**bb**) and thus, unlike their parents, express the recessive phenotype.

This is what Mendel found when he crossed monohybrids. It occurs because meiosis separates the two alleles of each heterozygous parent so that 50% of the gametes will carry one allele and 50% the other and when the gametes are brought together at random, each **B** (or **b**)-carrying egg will have a 1 in 2 probability of being fertilized by a sperm carrying **B** (or **b**). (Left table)

Results of random union of the two gametes produced by two individuals, each heterozygous for a given trait. As a result of meiosis, half the gametes produced by each parent with carry allele **B**; the other half allele **b**.			Results of random union of the gametes produced by an entire population with a gene pool containing 80% **B** and 20% **b**.		
	0.5 **B**	0.5 **b**		0.8 **B**	0.2 **b**
0.5 **B**	0.25 **BB**	0.25 **Bb**	0.8 **B**	0.64 **BB**	0.16 **Bb**
0.5 **b**	0.25 **Bb**	0.25 **bb**	0.2 **b**	0.16 **Bb**	0.04 **bb**

However, the frequency of two alleles in an **entire population** of organisms is unlikely to be exactly the same. Let us take as a hypothetical case, a population of hamsters in which 80% of all the gametes in the population carry a dominant allele for black coat (**B**) and 20% carry the recessive allele for gray coat (**b**).

Random union of these gametes (right table) will produce a generation:

- 64% homozygous for **BB** (0.8 x 0.8 = 0.64)
- 32% **Bb** heterozygotes (0.8 x 0.2 x 2 = 0.32)
- 4% homozygous (**bb**) for gray coat (0.2 x 0.2 = 0.04)

So 96% of this generation will have black coats; only 4% gray coats.

Will gray coated hamsters eventually disappear? No. Let's see why not.

- All the gametes formed by **BB** hamsters will contain allele **B** as will one-half the gametes formed by heterozygous (**Bb**) hamsters.
- So, 80% (0.64 + .5*0.32) of the pool of gametes formed by this generation with contain **B**.
- All the gametes of the gray (**bb**) hamsters (4%) will contain **b** but one-half of the gametes of the heterozygous hamsters will as well.
- So 20% (0.04 + .5*0.32) of the gametes will contain **b**.

So we have duplicated the initial situation exactly. The proportion of allele **b** in the population has remained the same. The heterozygous hamsters ensure that each generation will contain 4% gray hamsters. Now let us look at an algebraic analysis of the same problem using the expansion of the binomial $(\mathbf{p}+\mathbf{q})^2$.

$$(p+q)^2 = p^2 + 2pq + q^2 \tag{20.2.1}$$

The total number of genes in a population is its *gene pool*.

- Let p represent the frequency of one gene in the pool and q the frequency of its single allele.
- So, $p + q = 1$
  - p^2 = the fraction of the population homozygous for p
  - q^2 = the fraction homozygous for q
  - $2pq$ = the fraction of heterozygotes
- In our example, **p** = 0.8, **q** = 0.2, and thus

$$(0.8+0.2)^2 = (0.8)^2 + 2(0.8)(0.2) + (0.2)^2 = 064 + 0.32 + 0.04 \tag{20.2.2}$$

The algebraic method enables us to work backward as well as forward. In fact, because we chose to make **B** fully dominant, the only way that the frequency of **B** and **b** in the gene pool could be known is by determining the

frequency of the recessive phenotype (gray) and computing from it the value of **q**.

$\mathbf{q}^2$ = 0.04, so **q** = 0.2, the frequency of the **b** allele in the gene pool. Since **p** + **q** = 1, **p** = 0.8 and allele **B** makes up 80% of the gene pool. Because **B** is completely dominant over **b**, we cannot distinguish the **Bb** hamsters from the **BB** ones by their phenotype. But substituting in the middle term (**2pq**) of the expansion gives the percentage of heterozygous hamsters. **2pq** = (2)(0.8)(0.2) = 0.32

So, recessive genes do not tend to be lost from a population no matter how small their representation.

> **Hardy-Weinberg law**
>
> So long as certain conditions are met (discussed below), **gene frequencies** and **genotype ratios** in a randomly-breeding population remain constant from generation to generation. This is known as the Hardy-Weinberg law.

The Hardy-Weinberg law is named in honor of the two men who first realized the significance of the binomial expansion to population genetics and hence to evolution. Evolution involves changes in the gene pool. A population in Hardy-Weinberg equilibrium shows no change. What the law tells us is that populations are able to maintain a reservoir of variability so that if future conditions require it, the gene pool can change. If recessive alleles were continually tending to disappear, the population would soon become homozygous. Under Hardy-Weinberg conditions, genes that have no present selective value will nonetheless be retained.

When the Hardy-Weinberg Law Fails

To see what forces lead to evolutionary change, we must examine the circumstances in which the Hardy-Weinberg law may fail to apply. There are five:

1. mutation
2. gene flow
3. genetic drift
4. nonrandom mating
5. natural selection

Mutation

The frequency of gene **B** and its allele **b** will not remain in Hardy-Weinberg equilibrium if the rate of mutation of **B** -> **b** (or vice versa) changes. By itself, this type of mutation probably plays only a minor role in evolution; the rates are simply too low. However, gene (and whole genome) duplication - a form of mutation - probably has played a major role in evolution. In any case, evolution absolutely depends on mutations because this is the only way that new alleles are created. After being shuffled in various combinations with the rest of the gene pool, these provide the raw material on which natural selection can act.

Gene Flow

Many species are made up of local populations whose members tend to breed within the group. Each local population can develop a gene pool distinct from that of other local populations. However, members of one population may breed with occasional immigrants from an adjacent population of the same species. This can introduce new genes or alter existing gene frequencies in the residents.

In many plants and some animals, gene flow can occur not only between subpopulations of the same species but also between different (but still related) species. This is called **hybridization**. If the hybrids later breed with one of the parental types, new genes are passed into the gene pool of that parent population. This process is called **introgression**. It is simply gene flow between species rather than within them.

Comparison of the genomes of contemporary humans with the genome recovered from Neanderthal remains shows that from 1–3% of our genes were acquired by introgression following mating between members of the two populations tens of thousands of years ago.

Whether within a species or between species, gene flow increases the variability of the gene pool.

Genetic Drift

As we have seen, interbreeding often is limited to the members of local populations. If the population is small, Hardy-Weinberg may be violated. Chance alone may eliminate certain members out of proportion to their numbers in the population. In such cases, the frequency of an allele may begin to drift toward higher or lower values. Ultimately, the allele may represent 100% of the gene pool or, just as likely, disappear from it.

Drift produces evolutionary change, but there is no guarantee that the new population will be more fit than the

original one. Evolution by drift is aimless, not adaptive.

Nonrandom Mating

One of the cornerstones of the Hardy-Weinberg equilibrium is that mating in the population must be random. If individuals (usually females) are choosy in their selection of mates, the gene frequencies may become altered. Darwin called this **sexual selection**.

Nonrandom mating seems to be quite common. Breeding territories, courtship displays, "pecking orders" can all lead to it. In each case certain individuals do not get to make their proportionate contribution to the next generation.

Assortative mating

Humans seldom mate at random preferring phenotypes like themselves (e.g., size, age, ethnicity). This is called *assortative mating*. Marriage between close relatives is a special case of assortative mating. The closer the kinship, the more alleles shared and the greater the degree of **inbreeding**. Inbreeding can alter the gene pool. This is because it predisposes to **homozygosity**. Potentially harmful recessive alleles — invisible in the parents — become exposed to the forces of natural selection in the children.

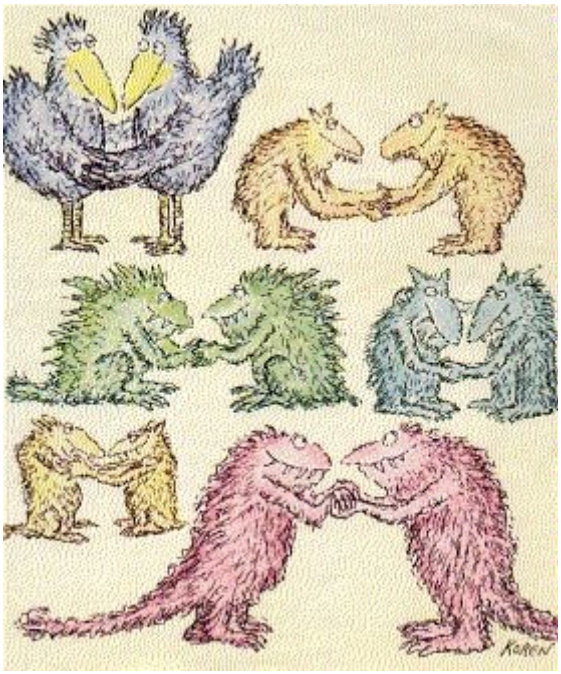

Figure 18.6.1: Assortative mating. (Drawing by Koren © 1977 The New Yorker Magazine, Inc.)

It turns out that many species - plants as well as animals - have mechanisms be which they avoid inbreeding. Examples:

- Link to discussion of self-incompatibility in plants.
- Male mice use olfactory cues to discriminate against close relatives when selecting mates. The preference is learned in infancy - an example of imprinting. The distinguishing odors are controlled by the MHC alleles of the mice and are detected by the vomeronasal organ (VNO).

Natural Selection

If individuals having certain genes are better able to produce mature offspring than those without them, the frequency of those genes will increase. This is simply expressing Darwin's natural selection in terms of alterations in the gene pool. (Darwin knew nothing of genes.) Natural selection results from *differential mortality* and/or *differential fecundity*.

Mortality Selection

Certain genotypes are less successful than others in surviving through to the end of their reproductive period. The evolutionary impact of mortality selection can be felt anytime from the formation of a new zygote to the end (if there is one) of the organism's period of fertility. Mortality selection is simply another way of describing Darwin's criteria of fitness: **survival**.

Fecundity Selection

Certain phenotypes (thus genotypes) may make a disproportionate contribution to the gene pool of the next generation by producing a disproportionate number of young. Such fecundity selection is another way of

describing another criterion of fitness described by Darwin: **family size**. In each of these examples of natural selection, certain phenotypes are better able than others to contribute their genes to the next generation. Thus, by Darwin's standards, they are more **fit**. The outcome is a gradual change in the gene frequencies in that population.

Calculating the Effect of Natural Selection on Gene Frequencies

The effect of natural selection on gene frequencies can be quantified. Let us assume a population containing

- 36% homozygous dominants (**AA**)
- 48% heterozygotes (**Aa**) and
- 16% homozygous recessives (**aa**)

The gene frequencies in this population are $p = 0.6$ and $q = 0.4$. The heterozygotes are just as successful at reproducing themselves as the homozygous dominants, but the homozygous recessives are only 80% as successful. That is, for every 100 **AA** (or **Aa**) individuals that reproduce successfully only 80 of the **aa** individuals succeed in doing so. The *fitness* (w) of the recessive phenotype is thus 80% or 0.8.

Their relative disadvantage can also be expressed as a *selection coefficient*, s, where

$$s = 1 - w \tag{20.2.3}$$

In this case,

$$s = 1 - 0.8 = 0.2. \tag{20.2.4}$$

The change in frequency of the dominant allele (Δp) after one generation is expressed by the equation

$$\Delta p = \frac{s p_0 q_0^2}{1 - s q_0^2} \tag{20.2.5}$$

where p_0 and q_0 are the initial frequencies of the dominant and recessive alleles respectively. Substituting, we get

$$\Delta p = \frac{(0.2)(0.6)(0.4)^2}{1 - (0.2)(0.4)^2} \tag{20.2.6}$$

$$= \frac{0.019}{0.968} \tag{20.2.7}$$

$$= 0.02 \tag{20.2.8}$$

So, in one generation, the frequency of allele **A** rises from its initial value of 0.6 to 0.62 and that of allele **a** declines from 0.4 to 0.38 ($q = 1 - p$).

The new equilibrium produces a population of

- 38.4% homozygous dominants (an increase of 2.4%) ($\mathbf{p^2}$ = 0.384)
- 47.1% heterozygotes (a decline of 0.9%)(**2pq** = 0.471) and
- 14.4% homozygous recessives (a decline of 1.6%)($\mathbf{q^2}$ = 0.144)

If the fitness of the homozygous recessives continues unchanged, the calculations can be reiterated for any number of generations. If you do so, you will find that although the frequency of the recessive genotype declines, the **rate** at which **a** is removed from the gene pool declines; that is, the process becomes less efficient at purging allele **a**. This is because when present in the heterozygote, **a** is protected from the effects of selection.
