

### **Cystic Fibrosis (Example of Hardy-Weinberg Equilibrium):**

The probability is  $p = 0.02$  in a Caucasian population.

Therefore,  $q = 0.98$

The frequency of heterozygotes is  $2pq = 2 * (.02) * (.98) = .0392 \sim .04$

The frequency of recessive homozygotes (suffer from cystic fibrosis) is  $p^2$ , or  $(.02) * (.02) = 0.0004$

What does this tell us? Compare the probabilities. In general, recessive alleles are more likely to be present in heterozygous individuals than in homozygous individuals.

## **Deviations from Hardy-Weinberg Equilibrium**

### **Inbreeding - How does it affect a population?**

Inbreeding (when two relatives produce offspring) changes genotype proportions. Since the inbreeders are related, it's more likely than random mating that for one characteristic they will have matching alleles.

This includes recessive alleles. Therefore, the frequency of homozygotes increases (as the genes the offspring receives from both related parents are more likely to be the same) and the frequency of heterozygotes decreases. Consider the interaction of the following three cases:

AA \* AA —————> Homozygote frequency increases (AA)

AB \* AB —————> Heterozygote frequency decreases (AB)

BB \* BB —————> Homozygote frequency increases (BB)

### **Small Population Sizes: Genetic Drift**

In a small population, the sampling of gametes and fertilization to create zygotes causes random error in allele frequencies. This results in a deviation from the Hardy-Weinberg Equilibrium. This deviation is larger at small sample sizes and smaller at large sample sizes. Think of it like tossing coins - the average result for tossing two coins might be 100% heads. The average for tossing four coins might be 75% heads. But if you take a sample of 10,000 coin tosses, then you would have a 50% probability for heads. The direction of this change is random: the dominant or recessive allele might be over or under represented in the next generation relative to the predicted HWE values.

This effect is called **genetic drift**, or that the amplitude of allele frequency fluctuation from one generation to the next increases in small populations. In a small population, genetic drift can result in a loss of variety across the entire genome over time. This even can result in a loss of **polymorphism** (alternate alleles) and driving the frequency of one allele to 1.

How is this relevant to evolution? The fluctuation of allele frequencies in a small isolated population might lead to novel genetic combinations that would not be possibly merely through selection. There are several different situations, which are described below. Alternatively, genetic drift may just reduce genetic diversity (**evolutionary potential**).

### **Small Population Sizes: Population Bottlenecks**

A **population bottleneck** occurs when a population undergoes a severe decrease in size. The effect of genetic drift on this new population is much higher than on the previous population. The new population will be much reduced in genetic diversity: consider the diagram below. Of a population with equal proportions of blue, yellow, and white marbles amongst a population numbering in the 100s, the new population has 5 blue marbles, 1 white marble, and no yellow marbles. This rapid change in allele frequencies could lead to divergence and a new species forming.

Note that a population bottleneck does not involve migration (that's the next topic). The most likely causes of a population bottleneck are disease, habitat loss, overharvesting leading to insufficient resources, or climate change.

An example is the prairie chickens of the Illinois plains. Their habitat loss resulted in many smaller populations, rather than a large unified population. The smaller population sizes led to a decrease in diversity and a decrease in the viability (i.e. the number of hatching chicks).

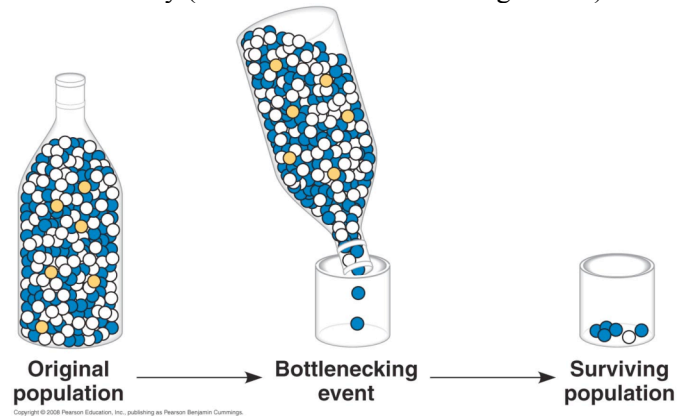


Figure 1: Population Bottlenecks

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### Small Population Sizes: Founder Events

In this case, a small population of a species moves to a new habitat. The effects are mostly similar to population bottlenecks (reduced genetic diversity in the new population, rapid change in allele frequency, high potential for divergence and speciation) with the exception that there would be more selection occurring due to the new habitat.

An example would be human colonization. African *Homo sapiens* have the most genetic diversity of any human population, while Europe, Asia, and the Americas have lower genetic diversity. This implies that the wellspring of the human species is in Africa (where the original population with high genetic diversity resided/resides), and the other populations are descendants of smaller migrating populations with lower genetic diversity. (This is what is termed the “Out of Africa” theory of human evolution.)

## Mutation

The dominant source of variation in the human genome. The process through which DNA is replicated in meiosis is not perfect, and it produces copy errors. There are an average of 1 in a billion nucleotide-copying errors per gamete per generation (the genome is 3 billion nucleotides long). Therefore, each time the human genome is replicated, it has an average of 3 mutations.

Mutations can arise in either a **somatic** cell line or in the **germ line**. Somatic cells are the cells that make up your body, so mutations in those cells are not passed on to the next generation. Mutations in the germ line, or in gametes, are passed on, so they are the mutations we talk about here.

These errors can be in one of four forms:

1. **Point mutation:** an alteration in the nucleotide coding sequence. For example, from a CTT/GAA sequence, a copy with a point-mutation might read CAT/GTA.
2. **Gene regulation:** a region controlling gene regulation and expression is altered.
3. **Gene copy number error:** a gene is copied more than it should be, so that the genome now has multiple copies of the same gene.
4. **Chromosome number and structure:** during meiosis, a gamete receives too many or two few chromosomes, or a chromosome is damaged. Examples include Trisomy 21 (Down’s syndrome) and plant polyploidy, when a plant can have nuclei with genetic material at 2N, 4N, 6N, etc...

**Note on basic genetic principles:** Definitions of basic genetic terms such as DNA, nucleotide, gene regulation, gene expression, chromosome, etc. were not and will not be covered in lecture. If this material is foreign, you can look them up in the textbook. In Campbell 8<sup>th</sup> edition, most of the material should be within chapters 15-18. Use the table of contents and the index to find whatever specific material you need clarified.