# EVOLUTION

# GENETIC CONSTANCY AND CREATION OF VARIABILITY

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# Biological Variations

- Biological variations may be due to
- Genotypic changes due to mutations and recombination, which produce permanent and heritable phenotypic alterations
- Environmentally induced changes, which produce temporary non-heritable phenotypic alterations

- **Phenotypes** are traits or characteristics of an organism that we can observe, such as size, color, shape, capabilities, behaviors, etc.
- Not all phenotypes can actually be seen. For example, blood types are phenotypes that we can only observe using laboratory techniques.
- Phenotypes can be caused by genes, environmental factors, or a combination of both.

- **Phenotypic variation**, then, is the variability in phenotypes that exists in a population.
- For example, people come in all shapes and sizes: height, weight, and body shape are phenotypes that vary.
- Hair, eye colour, and the ability to roll your tongue are variable phenotypes, too.
- All organisms can have phenotypic variation. In plants, flower colour and leaf shape are examples of variable phenotypes.

- **Phenotype**
- *Donax variabilis*

with diverse coloration and patterning in their phenotypes.

- When a characteristic or phenotype normally exists in a range or gradient, it **varies continuously**, like shades of gray as opposed to black and white. It's easy to think of examples of phenotypes that vary continuously, such as height and skin color. In between the shortest person in the world and the tallest person in the world, any height is possible, not just four feet, five feet, or six feet. And of course, skin comes in all kinds of shades, not just two or three.
- If you would make a frequency graph of the range of heights or skin colors in a group of people, it would look like a bell curve, with intermediate phenotypes being the most common. This is a good way to recognize continuous variation.

- In contrast, some phenotypes **vary discontinuously**. These phenotypes exist only at discrete intervals, like 'black and white' differences. For example, you can have blood type A, B, AB, or O, but there aren't any intermediate blood types in between. Another example is the ability to roll your tongue. Either you can or you can't, so this phenotype varies discontinuously.

- Environmentally caused variations may result from one factor or the combined effects of several factors, such as climate, food supply, and actions of other organisms.

(a) Caterpillars raised on a diet of oak flowers

(b) Caterpillars raised on a diet of oak leaves

# Genetic Variation

- Genetic variation can be described as the differences between organisms caused by alternate forms of DNA. Genetic variation in combination with *environmental variation* causes the total *phenotypic variation* seen in a population. The phenotypic variation is what is seen by the observer; the height of a plant for instance.

- The environmental variation is the difference in what each individual experiences. In our scenario, some plants may get more water and nutrients than others. If you remove the environmental differences from the overall variation, what you have left is the genetic variation. This variation represents the actual differences in DNA sequences between organisms.

# Causes of Genetic Variations

- **Mutations**
- **Gene Flow**
- **Genetic recombination**
- **Multiple alleles**

# Causes of genetic variation
# 1.Mutations

Any sudden change occurring in hereditary material is called as mutation

- They may be harmful, beneficial or neutral

# Gene Mutations

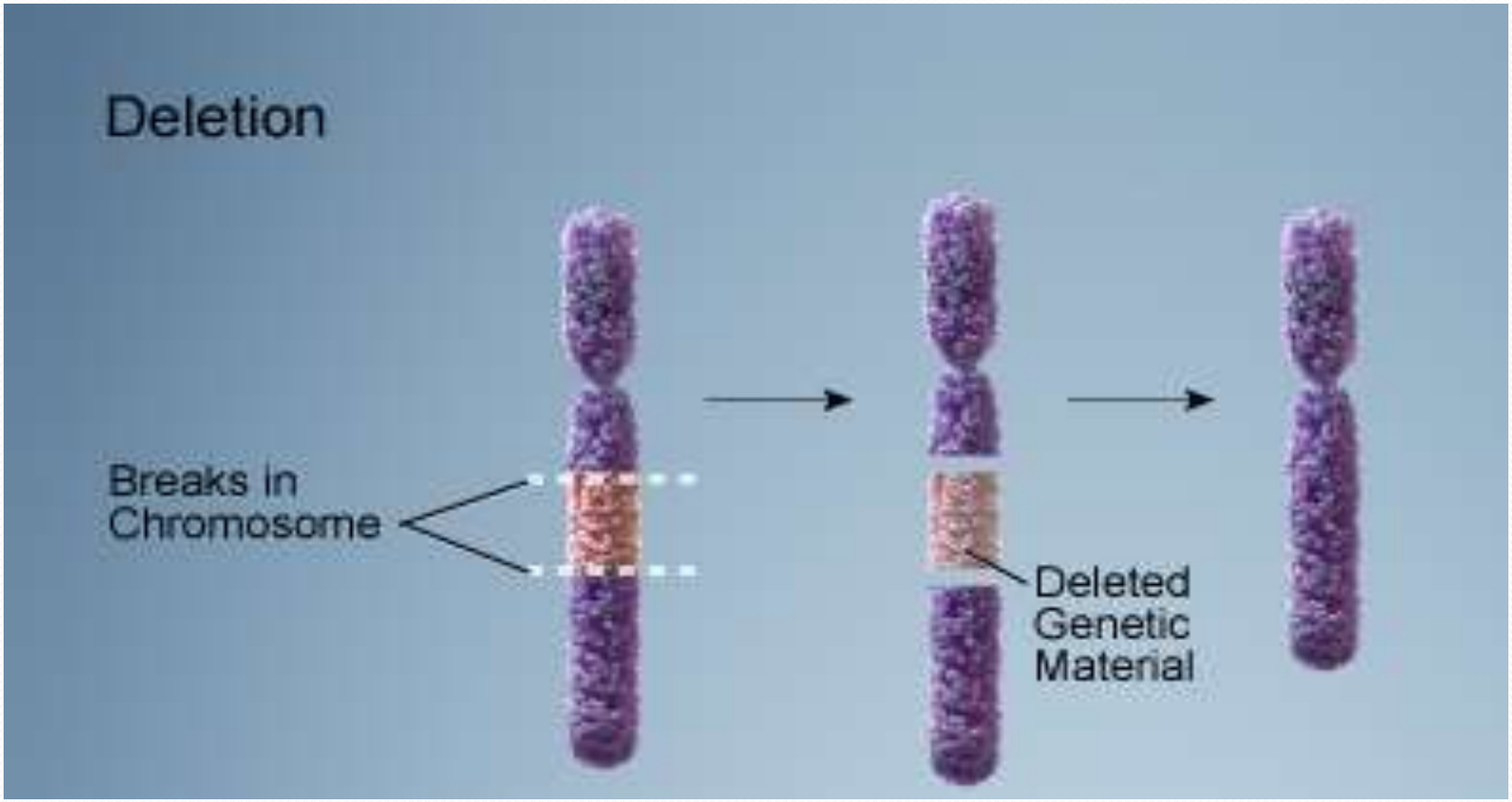
- Point mutations
- Intragenic mutations

# Chromosomal Mutations

- Any change in the **structure** or **number** of chromosomes
- Large scale: Affect *many* genes

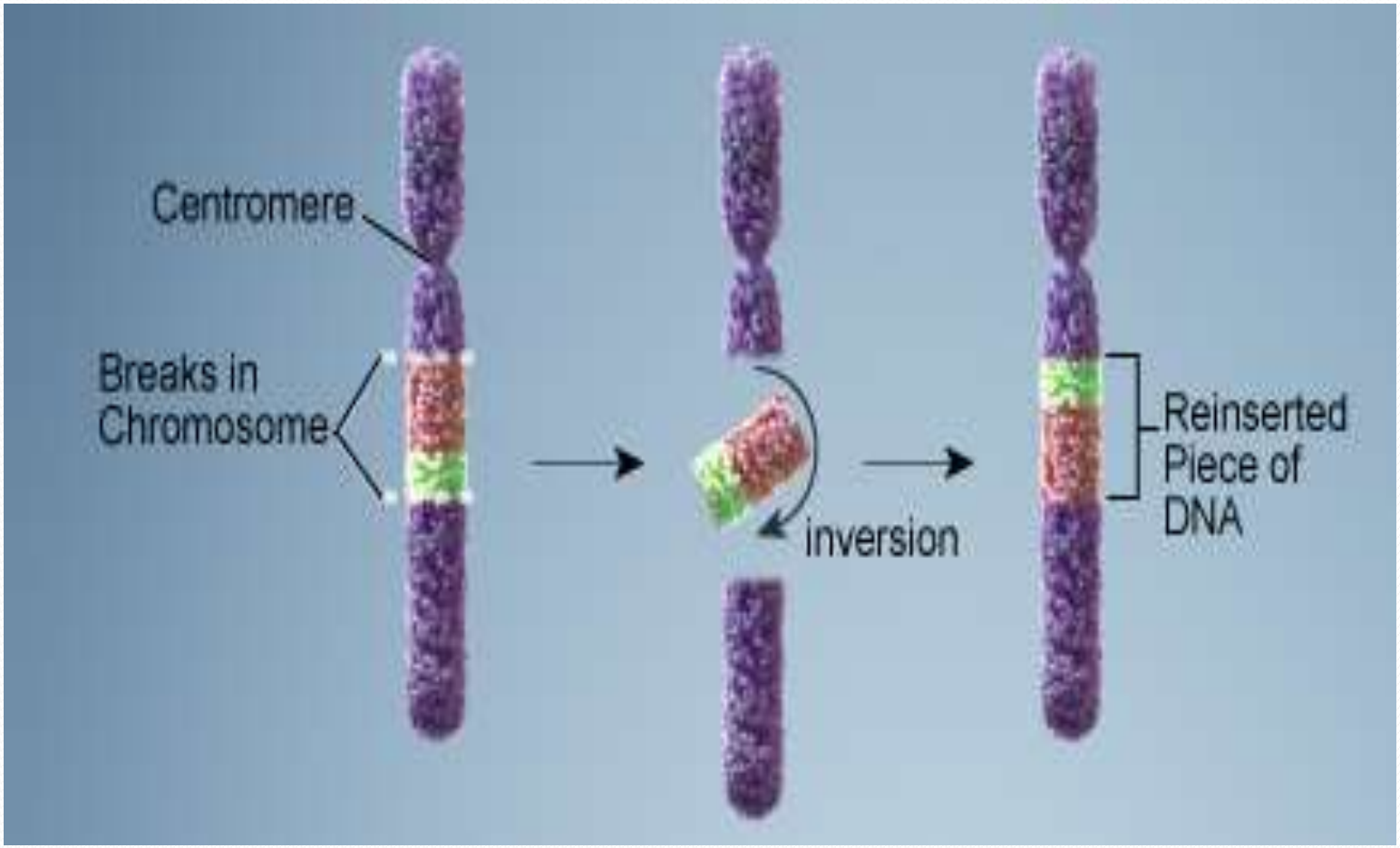
1. Deletion
2. Duplication
3. Inversion
4. Translocation

- Chromosomal Deletion



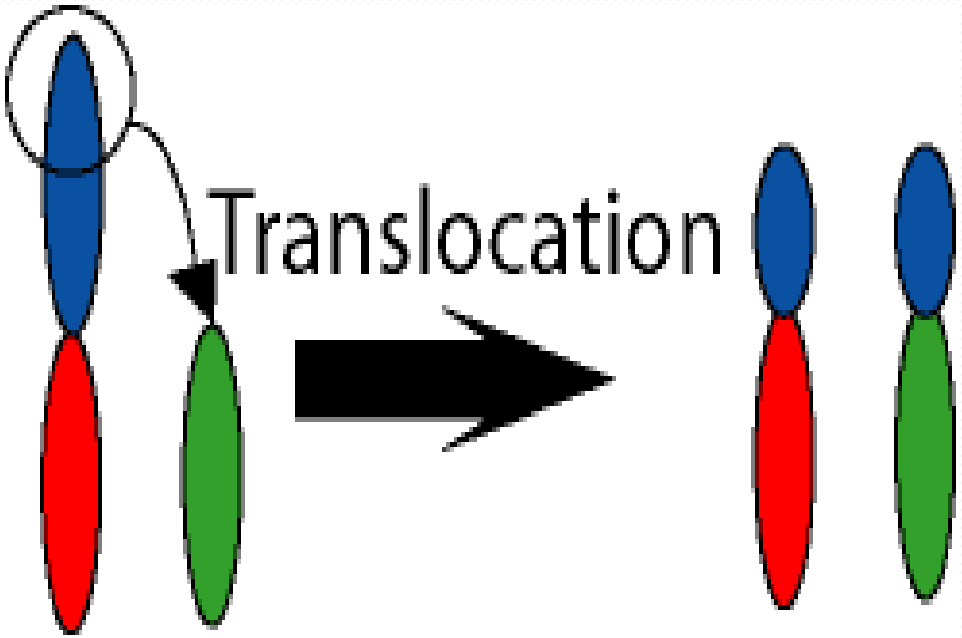

**One or more genes are removed**

- Chromosomal Inversion

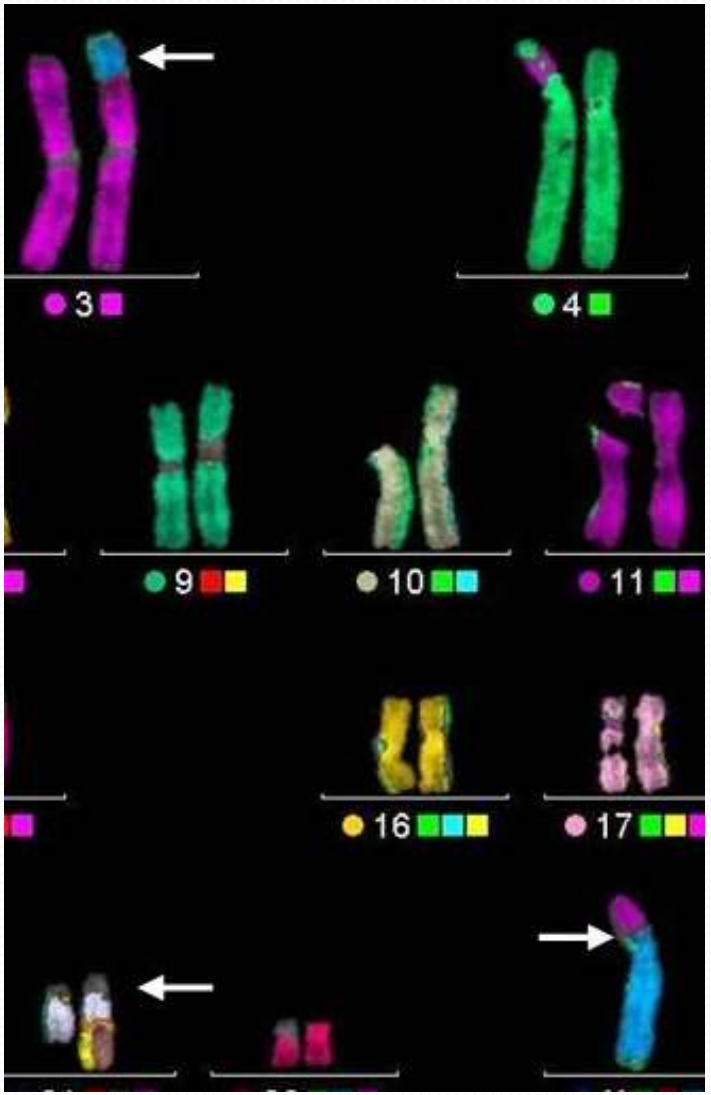



**a segment of genes flip end-to-end on the chromosome**

- Chromosomal Translocation

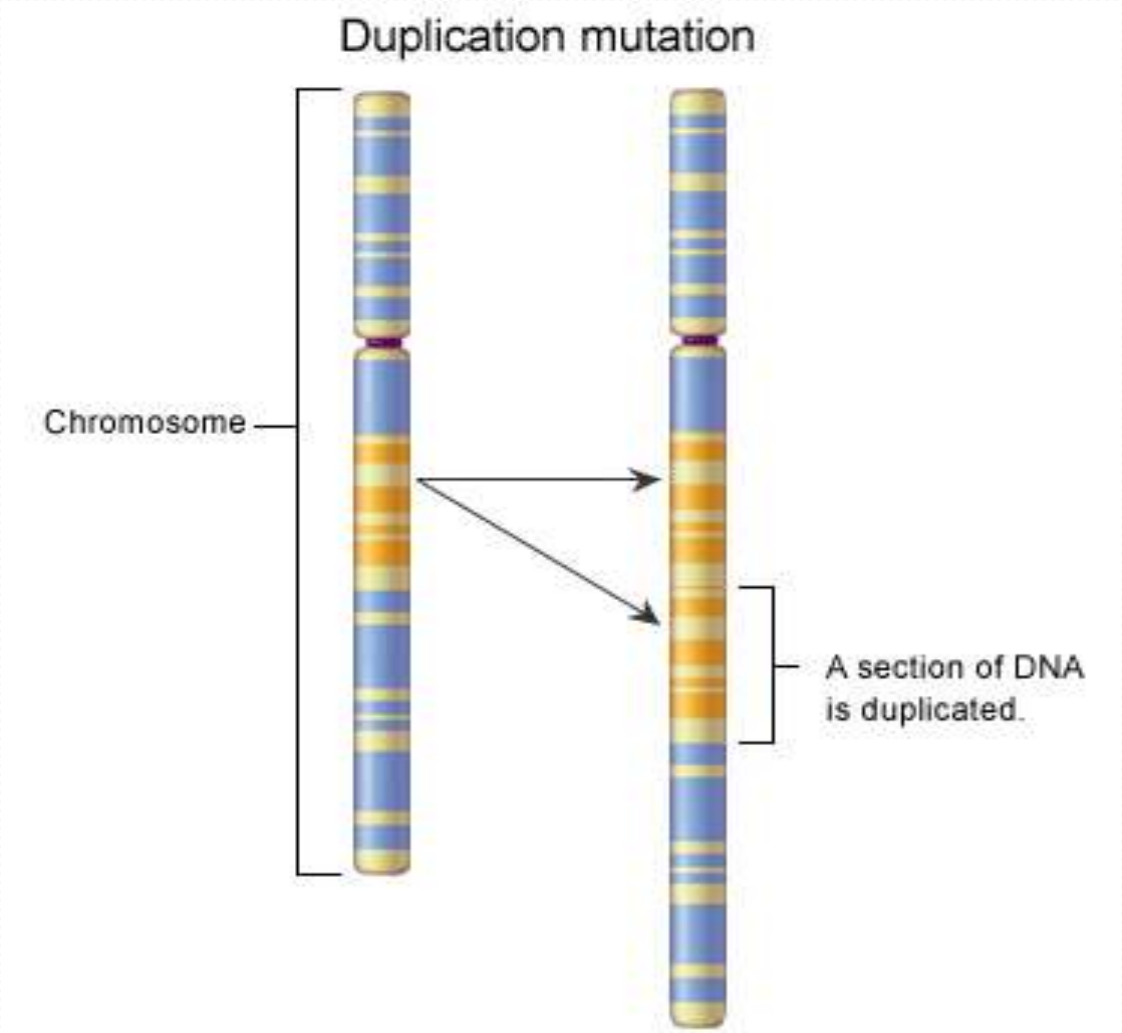



Material is swapped with another chromosome



# Duplication

- Occurs when a gene **sequence is repeated**



# Numerical Aberrations

- Numerical aberrations are generally caused by a failure in chromosome division during meiosis that results in gametic cells with an extra chromosome or a deficiency in the number of chromosomes. Variation in chromosome number involves
- **1) addition or loss of one or more chromosomes (Aneuploidy)**
- **2) addition or loss of one or more haploid sets of chromosomes (Euploidy)**

# 1- Aneuploidy ( Greek, aneu= uneven, ploids= units)

- When an organism gains or loses one or more chromosomes, but not a complete set, this condition is called aneuploidy. It leads to the variation in the number of chromosomes but not involves the whole set of chromosomes. The nuclei of aneuploids contain chromosomes whose number is not the true multiple of basic number (n).
- Examples: are **Down syndrome** (which has 47 chromosomes instead of 46) and **Turner syndrome** (45 chromosomes instead of 46). Here the number of chromosomes in the individual is not a true multiple of basic number n (n=23).

- Type of aneuploidy
- **i. Monosomy:** The loss of one chromosome produces a monosomic (2n-1) and the condition is known as monosomy.
- **ii. Trisomy:** The gain of one extra chromosome produces trisomic (2n+1) and the condition is called trisomy.
- **iii. Tetrasomy:** The gain of two extra chromosomes produces tetrasomic (2n+2) individuals and the condition is called Tetrasomy.
- **iv. Pentasomy:** The gain of three extra chromosomes produces pentasomic (2n+3) individuals and the condition is called Pentasomy.
- **V. Nullisomy:** It is a condition in which a pair of homologous chromosomes is completely lost.

# 2- Euploidy ( Greek,  ae= even or true,  ploids= units)

- When one or more complete haploid set of chromosomes are involved in the aberration, the resulting abnormality is called Euploidy. It is more tolerated in plants rather than animals. For example, if there is a human cell that has an extra set of 23 chromosomes it will have Euploidy.
- Types of Euploidy
- Ploidy refers to the number of homologous sets of chromosomes in the genome of a cell or an organism. Each set is designated by n.

- **i- Monoploidy:**
- Monoploidy: The state of having a single set of chromosomes is called monoploidy and is represented by 1n. The cell or organism with a single set of chromosomes is called a monoploid. Monoploidy is lethal in animals but in the case of plant species, this can be more tolerated.
- In most animal species this could mean death but there are few animal species where monoploidy is a normal part of the life cycle, such as **male wasps, ants, and bees.** The offsprings that have arisen from monoploidy are those that have developed from unfertilized eggs.

- **ii- polyploidy:**
- The condition in which a normally diploid cell or organism acquires one or more additional sets of chromosomes is called polyploidy. In other words, the polyploid cell or organism has three or more times the number of haploid chromosomes. Polyploidy arises as a result of the total nondisjunction of the chromosomes during mitosis or meiosis.

- **Polyploidy in plants:**
- Polyploidy is common among plants and has, in fact, been an important source of speciation in angiosperms. Particularly important is allopolyploidy, which involves the duplication of chromosomes in a hybrid plant.
- Typically, a diploid hybrid is sterile because it does not have the homologous chromosome pairs necessary for successful gamete formation during meiosis. However, in the case of tetra polyploids, the plant duplicates the set of chromosomes inherited from each parent, meiosis can occur, because each chromosome will have a homolog derived from its duplicate set. Thus, Tetrapolyploidy confers fertility on the previously sterile hybrid, which therefore attains the status of a complete species distinct from either parent.

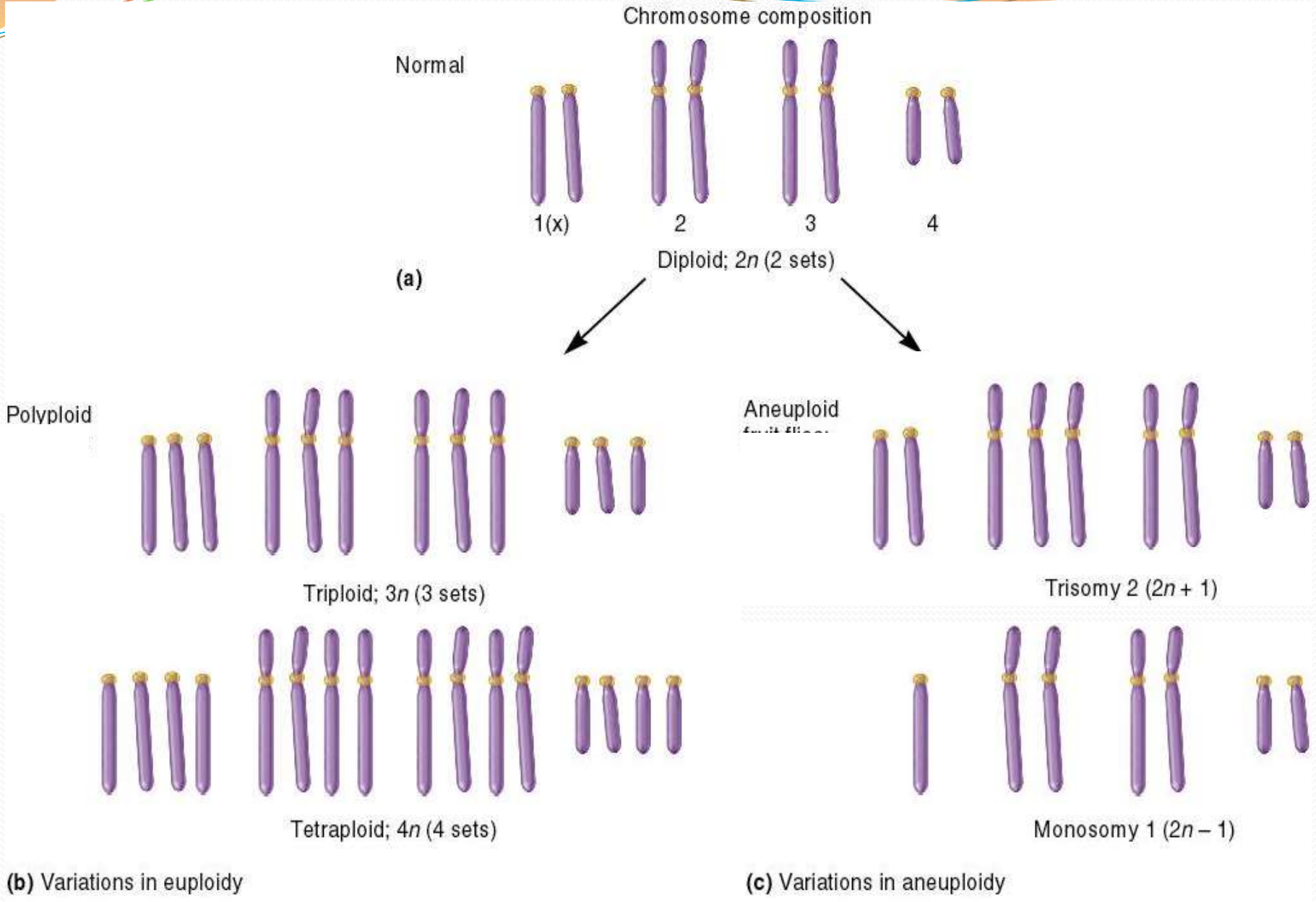
- Up to half of the known angiosperm species have been estimated to have arisen through polyploidy, including some of the species most prized by man. Plant breeders use this process, treating desirable hybrids with chemicals, such as colchicine, which are known to induce polyploidy.
- Polyploid animals are much less common, and the process appears to have had little effect on animal speciation.

## Tetraploid Plants (4n)

## Triploid Plants (3n)

# Benefit of Odd Ploidy-Induced Sterility

- Seedless fruit
  - watermelons and bananas
  - asexually propagated by human via cuttings
- Seedless flowers
  - Marigold flowering plants
- Prevention of cross pollination of transgenic plants


Chromosome composition
Normal
1(x)
2
3
4
Diploid; 2n (2 sets)
(a)
Polyploid
Triploid; 3n (3 sets)
Tetraploid; 4n (4 sets)
(b) Variations in euploidy
Aneuploid
Trisomy 2 (2n + 1)
Monosomy 1 (2n – 1)
(c) Variations in aneuploidy

- Mutation plays an important role in evolution. The ultimate source of all genetic variation is mutation. Mutation is important as the first step of evolution because it creates a new DNA sequence for a particular gene, creating a new allele. Recombination also can create a new DNA sequence (a new allele) for a specific gene through intragenic recombination. Mutation acting as an evolutionary force by itself has the potential to cause significant changes in allele frequencies over very long periods of time.

# Causes of Genetic Variation
# 2.Gene flow

- **Gene flow**, also called **gene migration**, the introduction of genetic material (by interbreeding) from one population of a species to another, thereby changing the composition of the gene pool of the receiving population. The introduction of new alleles through gene flow increases variability within the population and makes possible new combinations of traits. In humans gene flow usually comes about through the actual migration of human populations, either voluntary or forced.

- Although gene flow does not change allele frequencies for a species as a whole, it can alter allele frequencies in local populations. In the case of migration, the greater the difference in allele frequencies between the resident and the migrant individuals, and the larger the number of migrants, the greater the effect the migrants have in changing the genetic constitution of the resident population.

- Gene flow can also occur without migration.  When people travel to another area and successfully mate with people in the population there, a transfer of genes occurs between the populations even though the traveler returns home.

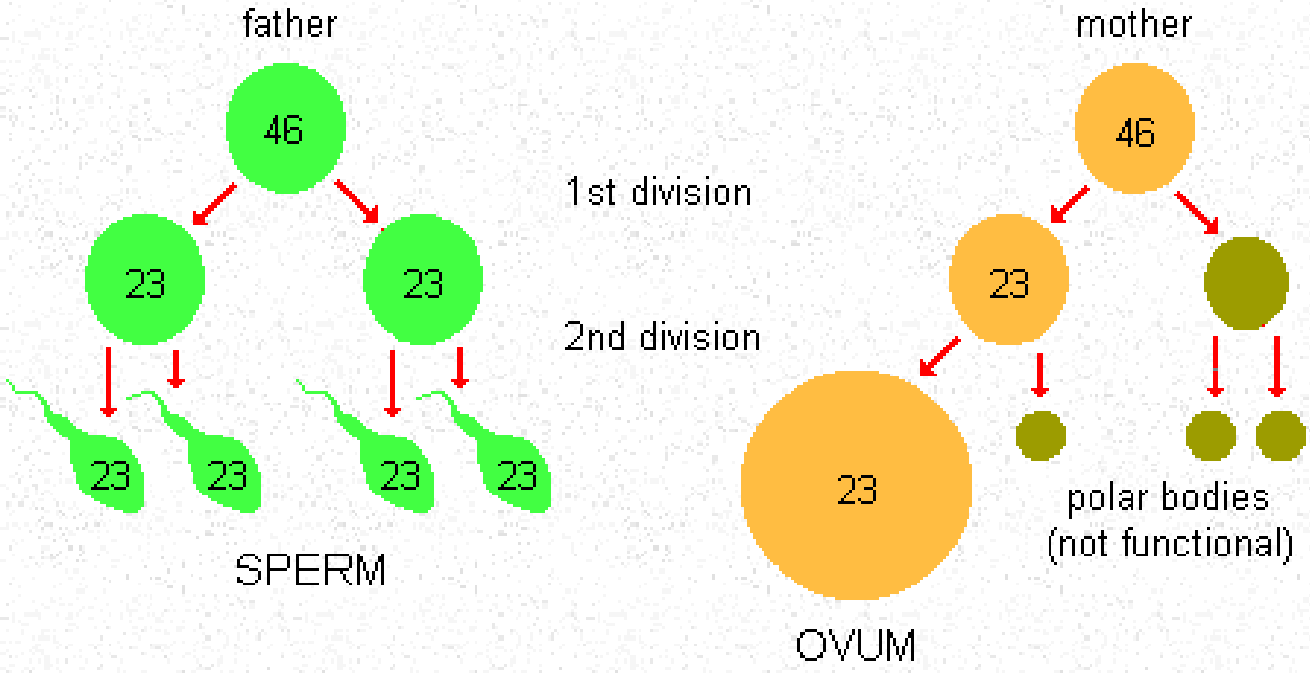
# Causes of Genetic Variation

## 3.Genetic Recombination

- Natural selection is usually the most powerful mechanism or process causing evolution to occur, however, it only selects among the existing variation already in a population. It does not create new genetic varieties or new combinations of varieties. One of the sources of those new combinations of genes is recombination during meiosis. It is responsible for producing genetic combinations not found in earlier generations.

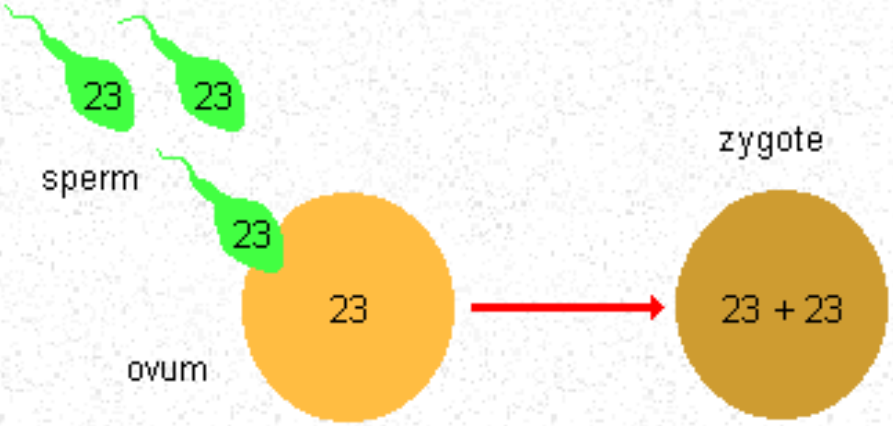
- Sperm and ova are radically different from somatic cells in the number of chromosomes that they contain. Both male and female sex cells normally get only half of the pair of parent chromosomes (23 for humans). Which half goes to any one sex cell is a matter of chance.
- Recombination by fertilization
- Recombination by meiosis
- Recombination by crossing over

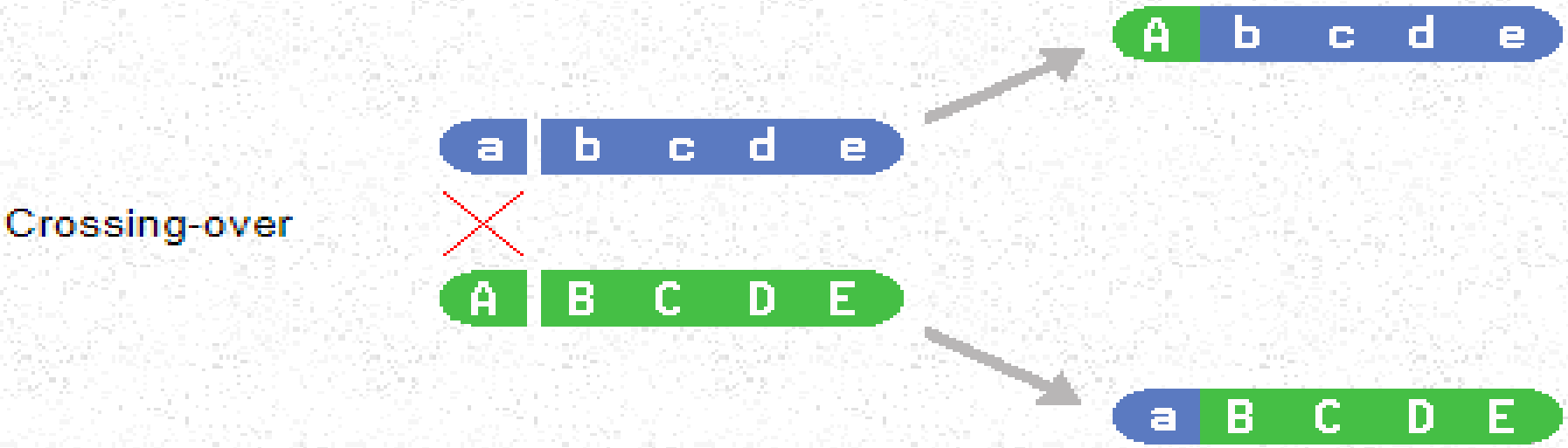
Net effect of the meiosis process in terms of chromosome numbers




- At conception, a single sperm and an ovum combine their chromosomes to produce a zygote with the normal full set of 46, but with a new combination of chromosomes distinct from either parent.

Sperm and ovum combining their chromosomes in a new zygote






- New combinations of existing genes are produced at the beginning of meiosis when the ends of chromosomes break and reattach, usually on their homologous chromosome. This crossing-over process results in an unlinking and recombination of parental genes. In the example below, one end of each chromosome of this homologous pair is exchanged along with the genes that they contain. The next generation inherits chromosomes with partially new sequences of alleles.

- The consequence of this recombination is the production of sperm and ova that can potentially add even greater diversity to a population's gene pool.  However, it does not result in new alleles.
- Subsequently, recombination by itself does not cause evolution to occur.
- Rather, it is a contributing mechanism that works with natural selection by creating combinations of genes that nature selects for or against.

# Causes of Genetic Variation
# 4.Multiple allelism

- A gene can have many different alleles within a population. A single organism can only have two alleles because it receives one from each of its parents. However, in a population, there can exist many different forms of a gene. Having multiple alleles for a gene leads to things like many different kinds of hair color and body size.
- One very important trait that is controlled by multiple alleles is Blood type. A, B and O are the three alleles and they can come together in different ways to make different blood types: AB, AA, BB, OO, AO, and BO.

- This concept of multiple alleles is important in the context of evolution. There has to be an ability for alleles to change so that natural selection can occur. Continued natural selection leads to evolution.
- Within a population, there a many different allelic forms that can control phenotypic traits like fur colour. In the above picture, each colouring pattern has an associated allele combination, and the mixing any of these breeds would produce a new allele combination.
- Many genes have more than two alleles (versions of the gene). This means that more than two forms of the allele exist in a species.

## Allele

| $C$ | $c^{ch}$ | $c^{h}$ | $c$ |
|---|---|---|---|

## Genotype

| $CC$ | $c^{ch}c^{ch}$ | $c^{h}c^{h}$ | $cc$ |
|---|---|---|---|

## Phenotype

| WILD TYPE: Brown fur | CHINCHILLA: Black-tipped white fur | HIMALAYAN: White fur with black paws, nose, ears, tail | ALBINO: White fur |
|---|---|---|---|
|  |  |  |  |

# Selection

- **Selection**, in biology, the preferential survival and reproduction or preferential elimination of individuals with certain genotypes (genetic compositions), by means of natural or artificial controlling factors.
- Natural selection can best be defined as **survival** and **reproduction** of the organisms that are genetically best adapted (suited) to the environment.

- The theory of evolution by natural selection was proposed by Charles Darwin and Alfred Russel Wallace in 1858. They argued that species with useful adaptations to the environment are more likely to survive and produce progeny than are those with less useful adaptations, thereby increasing the frequency with which useful adaptations occur over the generations.

- The limited resources available in an environment promotes competition in which organisms of the same or different species struggle to survive.
- In the competition for food, space, and mates that occurs, the less well-adapted individuals must die or fail to reproduce, and those who are better adapted do survive and reproduce.

- In the absence of competition between organisms, natural selection may be due to purely environmental factors, such as inclement weather or seasonal variations.
- Artificial selection , the selective agencies are those of human choice

# Natural Selection – High Reproductive Potential

- Darwin observed that organisms have the potential for very **high fertility**.
  - Organisms have the potential to produce, and often do produce large numbers of offspring.
  - Population size would quickly become unmanageable if all of the offspring survived.

- Natural Selection – Population Size Remains Constant
- Despite this high potential fertility, natural populations usually **remain constant in size**, except for small fluctuations.
  - Not all of the potential offspring survive.
  - Shows differential reproduction

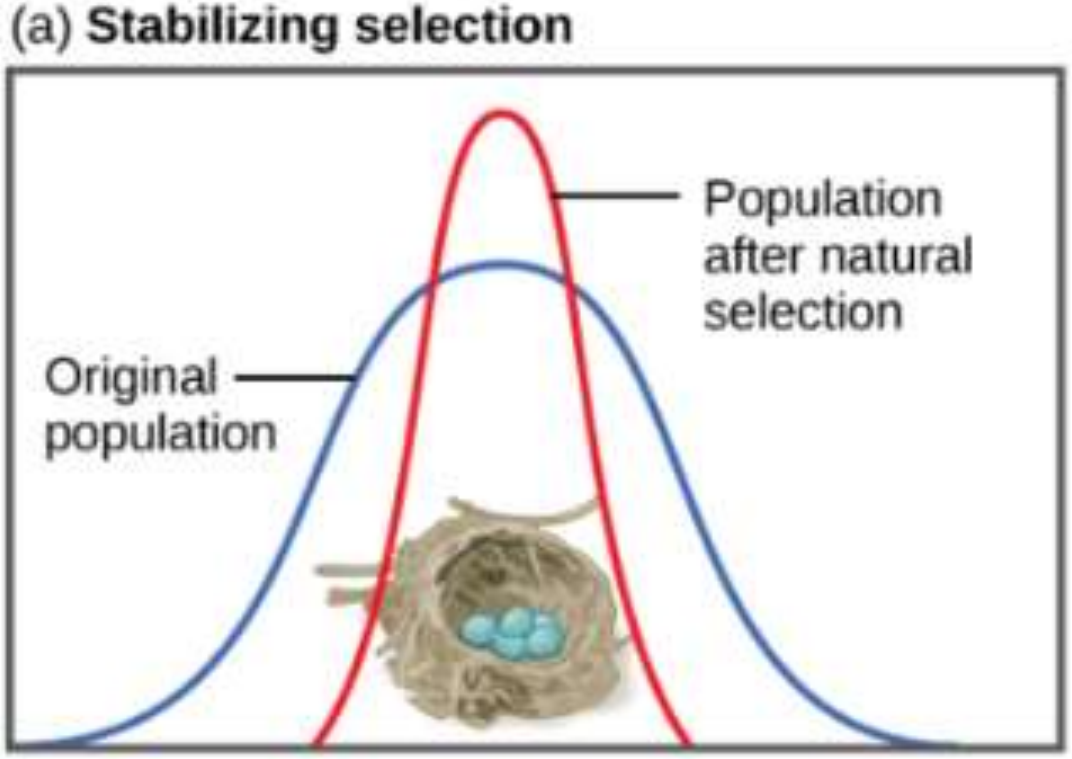
- Most mutations are deleterious with very low fitness. Such mutations will be removed quickly from the population by **negative selection or purifying selection**
- Mutations with higher fitness will be selected and allowed to multiply in the population by **positive selection.**
- Positive selection will lead to the eventual fixation of the selectively or preferentially favoured allele. Positive selection of quantitative characters is more often termed **directional selection**

# Types of Natural Selection

- When a population displays a normal distribution for a particular trait, natural selection can drive change in populations in different directions depending on the type of selection.
- **1. Stabilizing selection**
- **Stabilizing selection** results in a narrowing of the normal distribution, because individuals who had the 'average' phenotype, or the phenotype closest to the mean, tend to leave more offspring than those with phenotypes at either extreme.

- If natural selection favors an average phenotype by selecting against extreme variation, the population will undergo stabilizing selection. For example, in a population of mice that live in the woods, natural selection will tend to favor individuals that best blend in with the forest floor and are less likely to be spotted by predators. Assuming the ground is a fairly consistent shade of brown, those mice whose fur is most-closely matched to that color will most probably survive and reproduce, passing on their genes for their brown coat.

- Mice that carry alleles that make them slightly lighter or slightly darker will stand out against the ground and will more probably die from predation. As a result of this stabilizing selection, the population's genetic variance will decrease.

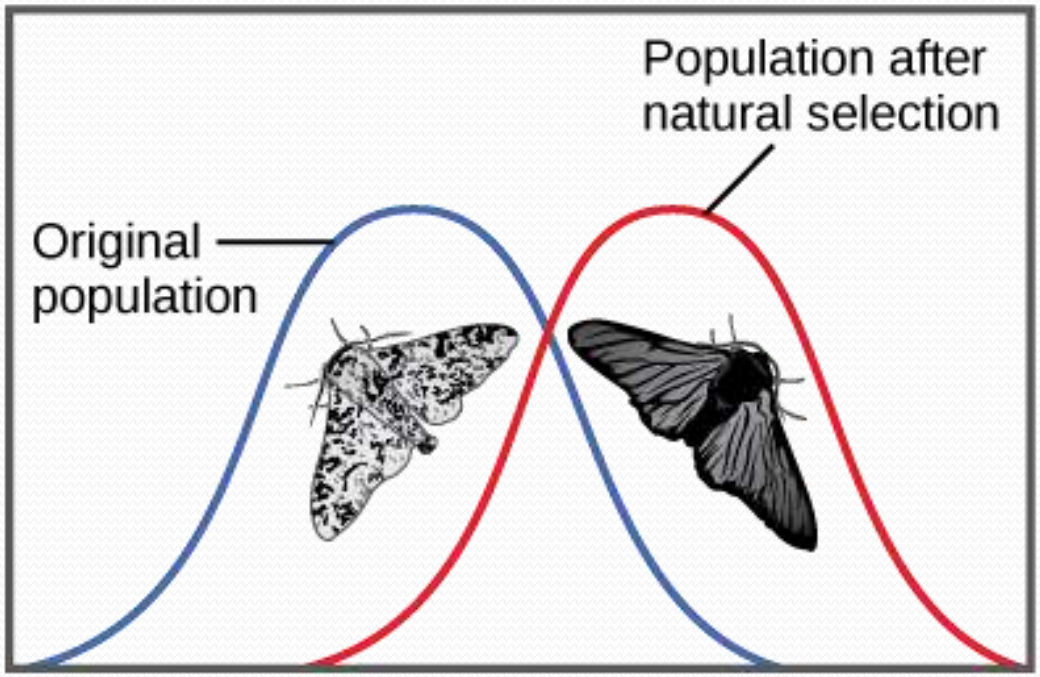



Robins typically lay four eggs, an example of stabilizing selection. Larger clutches may result in malnourished chicks, while smaller clutches may result in no viable offspring.

- **2. Directional selection** results in a shift toward one end of the normal distribution, because individuals who had one extreme of the phenotype tend to leave more offspring than those with the other extreme.
- When the environment changes, populations will often undergo directional selection, which selects for phenotypes at one end of the spectrum of existing variation.
- A classic example of this type of selection is the evolution of the peppered moth in eighteenth- and nineteenth-century England. Prior to the Industrial Revolution, the moths were predominately light in color, which allowed them to blend in with the light-colored trees and lichens in their environment. As soot began spewing from factories, the trees darkened and the light-colored moths became easier for predatory birds to spot.

- Over time, the frequency of the melanic form of the moth increased because their darker coloration provided camouflage against the sooty tree; they had a higher survival rate in habitats affected by air pollution. Similarly, the hypothetical mouse population may evolve to take on a different coloration if their forest floor habitat changed. The result of this type of selection is a shift in the population's genetic variance toward the new, fit phenotype.

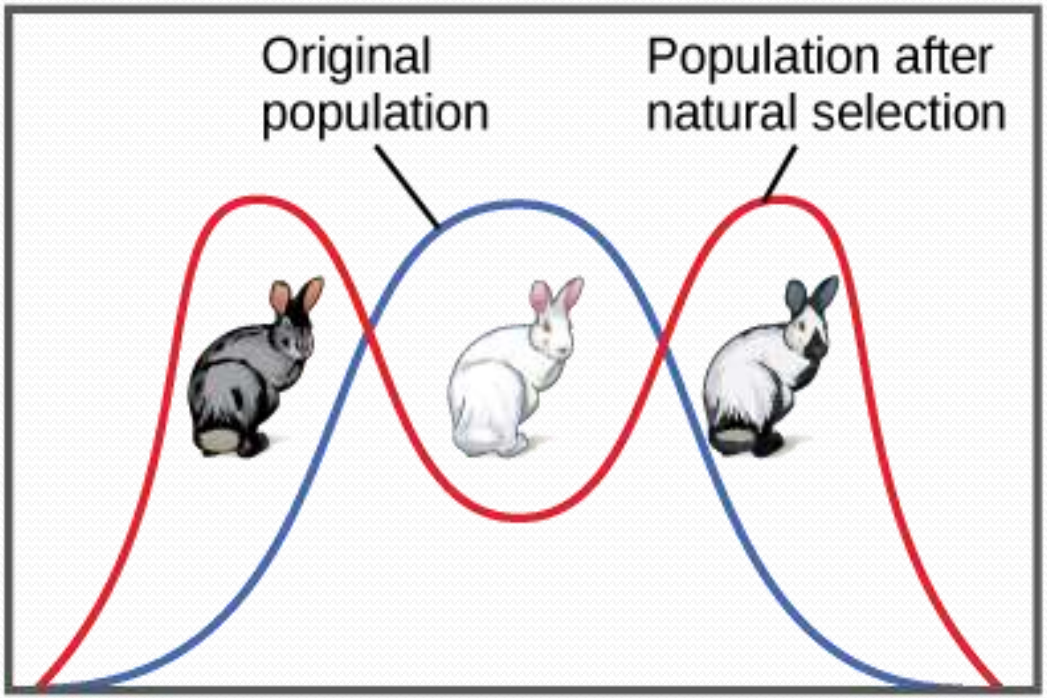
(b) **Directional selection**




Light-colored peppered moths are better camouflaged against a pristine environment; likewise, dark-colored peppered moths are better camouflaged against a sooty environment. Thus, as the Industrial Revolution progressed in nineteenth-century England, the color of the moth population shifted from light to dark, an example of directional selection.

- 3. **Disruptive or diversifying selection** results in separation of the normal distribution into two distributions with elimination of the middle of the peak, because individuals with either extreme phenotype tend to have more offspring than those with the intermediate phenotype.
- Sometimes natural selection can select for two or more distinct phenotypes that each have their advantages. In these cases, the intermediate phenotypes are often less fit than their extreme counterparts.

(c) **Diversifying selection**




In a hyphothetical population, gray and Himalayan (gray and white) rabbits are better able to blend with a rocky environment than white rabbits, resulting in diversifying selection.

# Genetic Drift

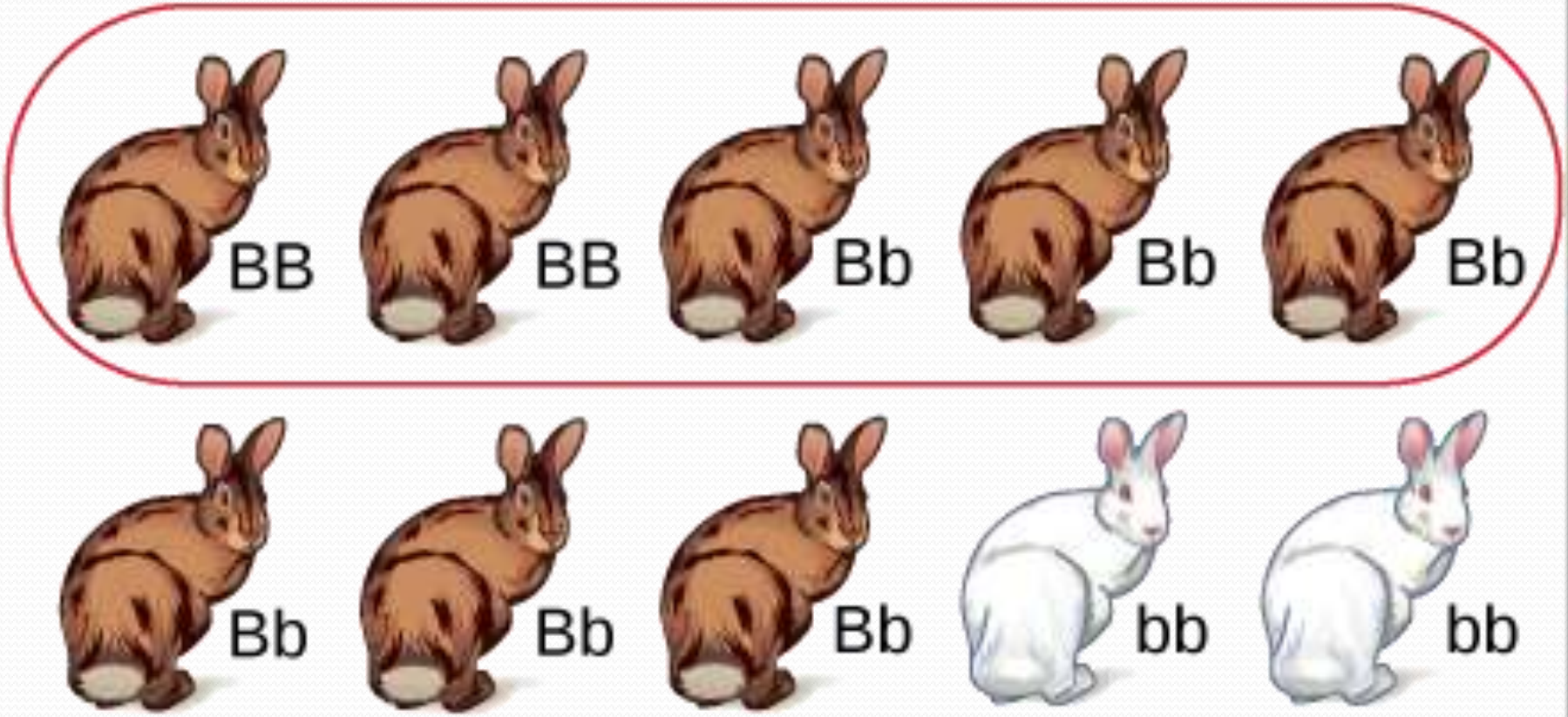
- **Genetic drift** is change in allele frequencies in a population from generation to generation that occurs due to chance events. Although genetic drift happens in populations of all sizes, its effects tend to be stronger in small populations.

- Let's make the idea of drift more concrete by looking at an example. As shown in the diagram below, we have a very small rabbit population that's made up of 888 brown individuals (genotype *BB* or *Bb*) and 222 white individuals (genotype *bb*). Initially, the frequencies of the *B* and *b* alleles are equal. What if, purely by chance, only the 555 circled individuals in the rabbit population reproduce? (Maybe the other rabbits died for reasons unrelated to their coat color, e.g., they happened to get caught in a hunter's snares.) In the surviving group, the frequency of the *B* allele is 0.7 and the frequency of the *b* allele is 0.3

# Genetic Drift

**First generation**

p (B gene frequency) = .5
q (b gene frequency) = .5

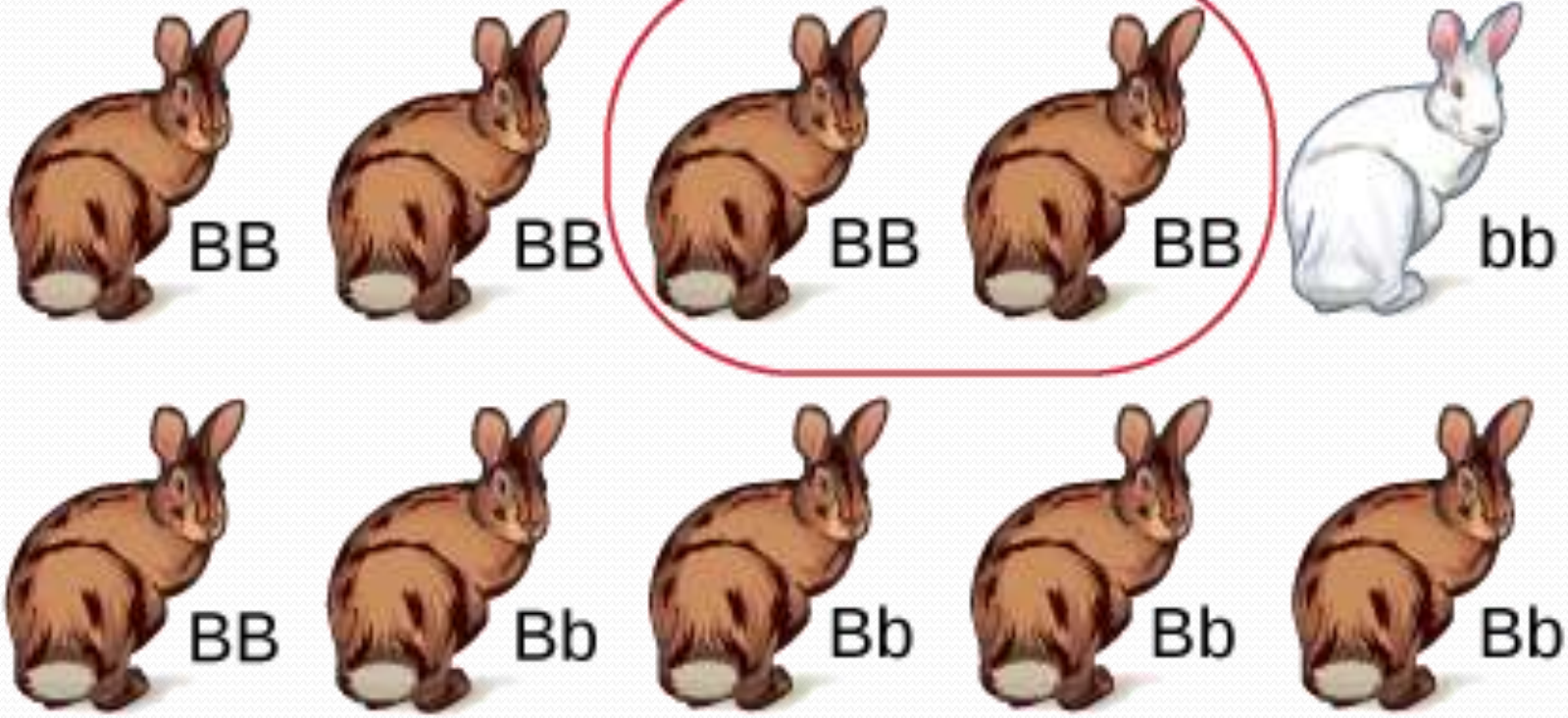


**5 rabbits reproduce**

## Second generation

p = .7

q = .3



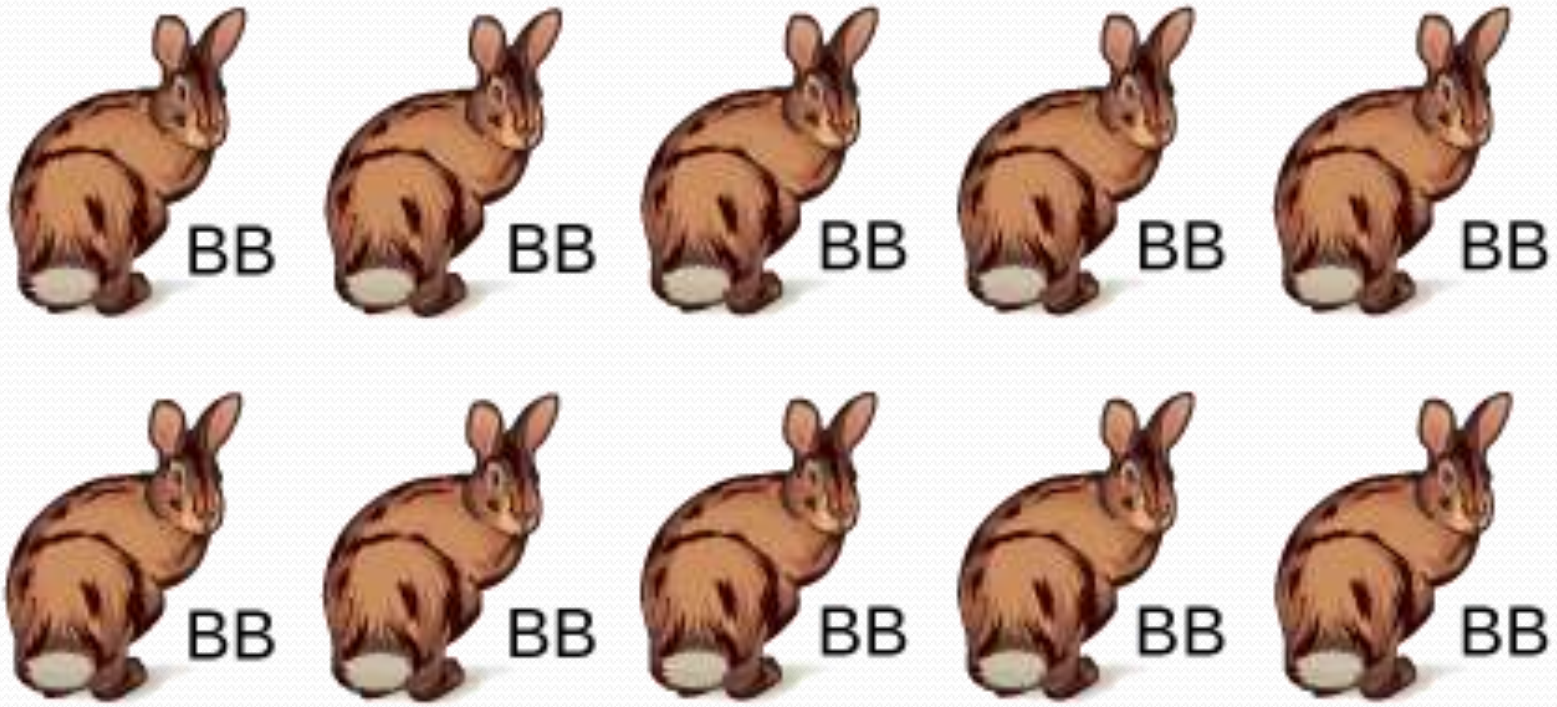
- In our example, the allele frequencies of the five lucky rabbits are perfectly represented in the second generation, as shown at right. Because the 555-rabbit "sample" in the previous generation had different allele frequencies than the population as a whole, frequencies of *B* and *b* in the population have shifted to 0.7 and 0.3 respectively.
- From this second generation, what if only two of the *BB* offspring survive and reproduce to yield the third generation? In this series of events, by the third generation, the *b* allele is completely lost from the population.

2 rabbits reproduce

## Third generation

p = 1

q = 0



- Larger populations are unlikely to change this quickly as a result of genetic drift. For instance, if we followed a population of 1000100001000 rabbits (instead of 101010), it's much less likely that the *b* allele would be lost (and that the *B* allele would reach 100% percent frequency, or **fixation**) after such a short period of time. If only half of the 1000100001000-rabbit population survived to reproduce, as in the first generation of the example above, the surviving rabbits (500500500 of them) would tend to be a much more accurate representation of the allele frequencies of the original population – simply because the sample would be so much larger.

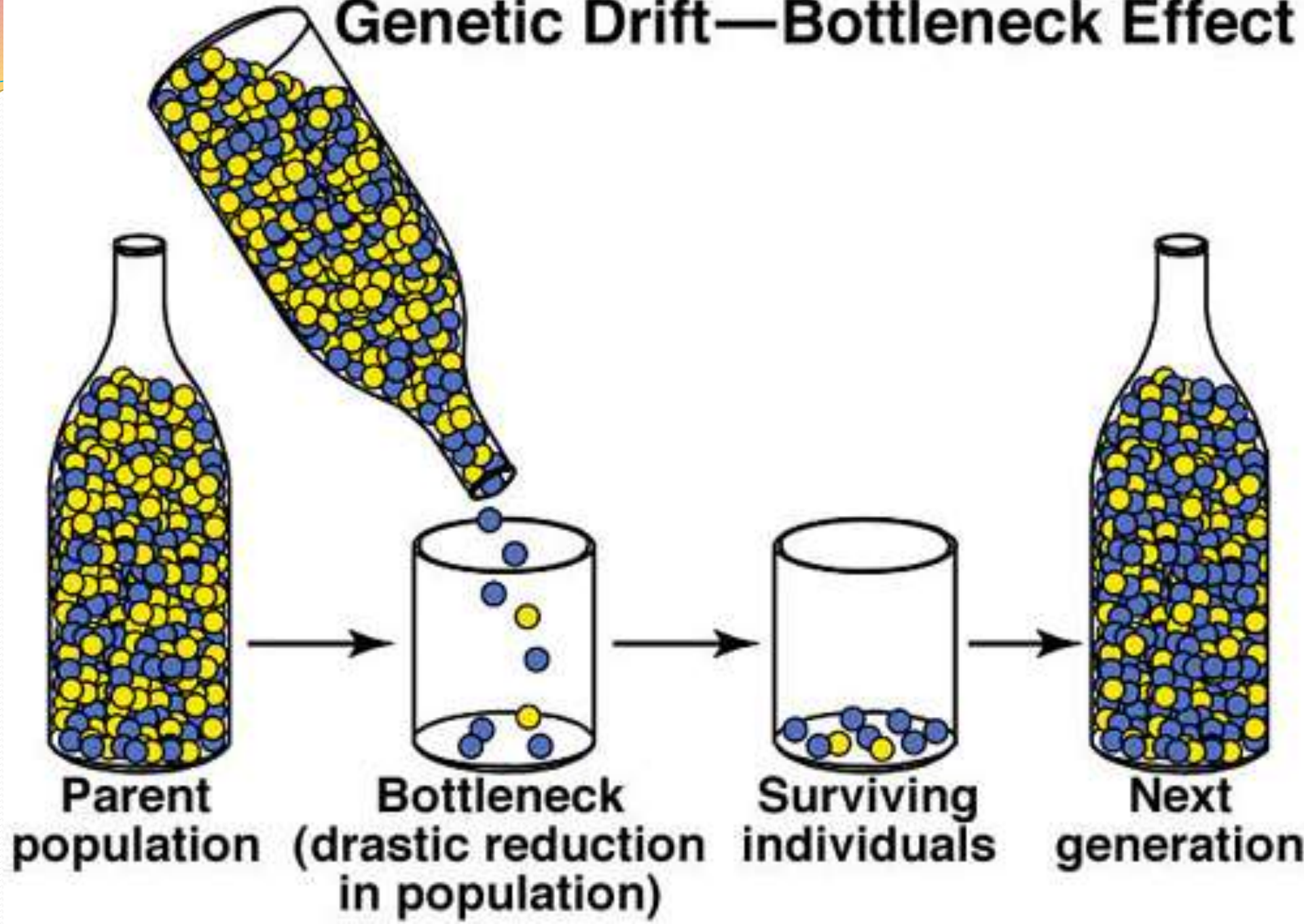
- Genetic drift, unlike natural selection, does not take into account an allele's benefit (or harm) to the individual that carries it. That is, a beneficial allele may be lost, or a slightly harmful allele may become fixed, purely by chance.
- A beneficial or harmful allele would be subject to selection as well as drift, but strong drift (for example, in a very small population) might still cause fixation of a harmful allele or loss of a beneficial one.

# The bottleneck effect

- The **bottleneck effect** is an extreme example of genetic drift that happens when the size of a population is severely reduced. Events like natural disasters (earthquakes, floods, fires) can decimate a population, killing most indviduals and leaving behind a small, random assortment of survivors.
- The allele frequencies in this group may be very different from those of the population prior to the event, and some alleles may be missing entirely. The smaller population will also be more susceptible to the effects of genetic drift for generations (until its numbers return to normal), potentially causing even more alleles to be lost.

- How can a bottleneck event reduce genetic diversity? Imagine a bottle filled with marbles, where the marbles represent the individuals in a population. If a bottleneck event occurs, a small, random assortment of individuals survive the event and pass through the bottleneck (and into the cup), while the vast majority of the population is killed off (remains in the bottle). The genetic composition of the random survivors is now the genetic composition of the entire population.

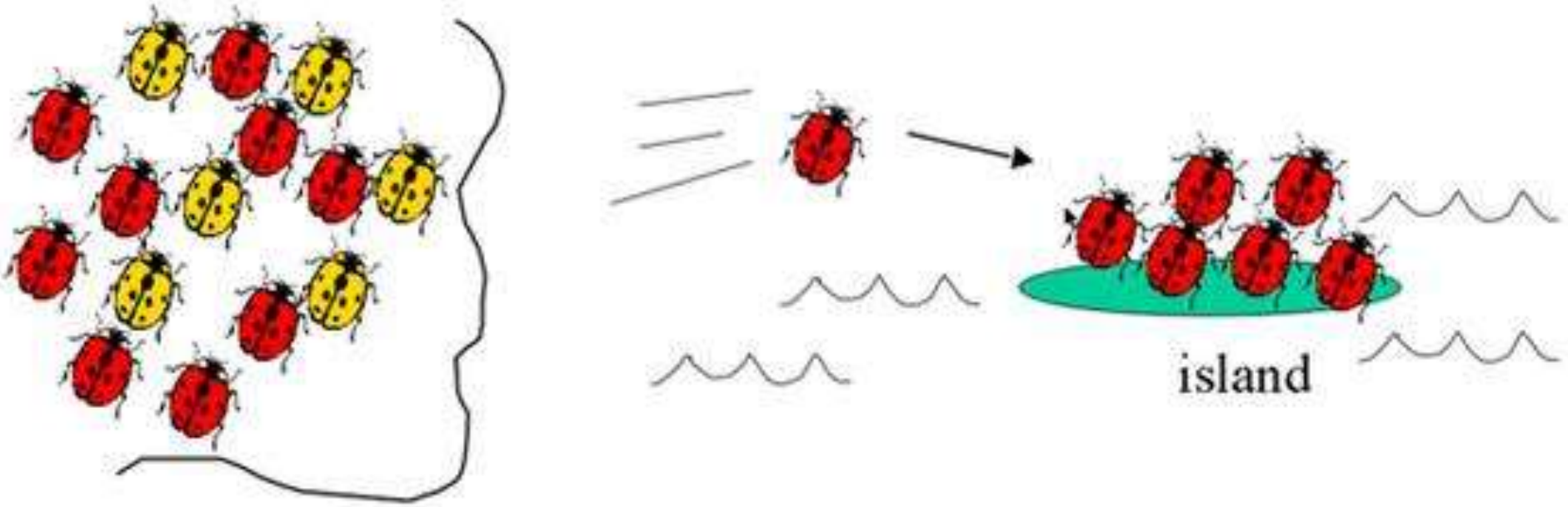
# Genetic Drift—Bottleneck Effect



# The founder effect

- The **founder effect** is another extreme example of drift, one that occurs when a small group of individuals breaks off from a larger population to establish a colony. The new colony is isolated from the original population, and the founding individuals may not represent the full genetic diversity of the original population. That is, alleles in the founding population may be present at different frequencies than in the original population, and some alleles may be missing altogether. The founder effect is similar in concept to the bottleneck effect, but it occurs via a different mechanism (colonization rather than catastrophe).

- **founder effect**: a few individuals from a population start a new population with a different allele frequency than the original population



- In the figure above, you can see a population made up of equal numbers of squares and circles. (Let's assume an individual's shape is determined by its alleles for a particular gene).
- Random groups that depart to establish new colonies are likely to contain different frequencies of squares and circles than the original population. So, the allele frequencies in the colonies (small circles) may be different relative to the original population. Also, the small size of the new colonies means they will experience strong genetic drift for generations.

# Genetic Polymorphism

- A **gene** is said to be **polymorphic** if more than one allele occupies that **gene's** locus within a population. For **example**, in dogs the E locus, can have any of five different alleles, known as E, $E^m$, $E^g$, $E^h$, and e. Varying combinations of these alleles contribute to the pigmentation and patterns seen in dog coats.
- There may be several **causes** of **polymorphism**: **polymorphism** can be maintained by a balance between variation created by new mutations and natural selection (see mutational load). **genetic** variation may be **caused** by frequency-dependent selection.

## Transient polymorphism

It is limited to the interim period of time during which one allele is replaced by a superior one. It is considered to be a by product of directional natural selection

## Balanced Polymorphism

It is relatively a permanent kind of equilibrium in which alleles are present in the population at steady state frequency.

## Neutral polymorphism

The action of the gene is neutral in its effect on the survival rate of the genotype which it contained.

# Thank You

This class prepared for

Fifth Semester BSc Botany Students

Little Flower College, Guruvayur

Affiliated to University of Calicut

Next Class:

Speciation