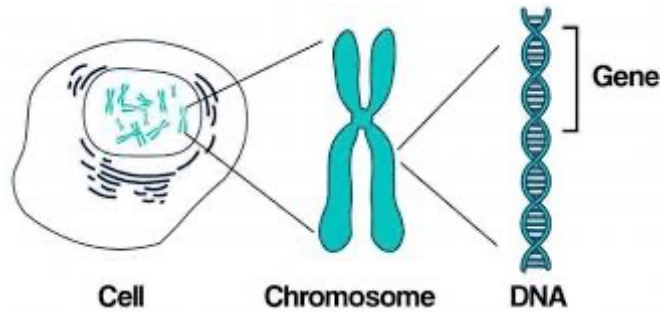


Population Genomics: Allele frequencies, genotype frequencies, genetic drift, gene flow, natural selection

@BeGenomics



DNA



↓ SNP

AAGGCCCCATGCAGCATCTGGAAGT

↓

Allele 1: AAGGCCCCATGCAGCATCTGGAAGT

Allele 2: AAGGCCCCATGCGGCATCTGGAAGT

A/A

G/G

A/G

Genotype Possible genotypes at this SNP:

- **A/A** (homozygous for A)
- **G/G** (homozygous for G)
- **A/G** (heterozygous, carrying both alleles)

Introduction to Allele Frequencies

- Genetic variation in populations can be analysed and quantified by the frequency of alleles
- Two fundamental calculations are central to population genetics: **allele frequencies** and **genotype frequencies**.
- Allele frequency is the proportion of a specific allele in a population.
- Allele frequencies are important in population genetics because they can change over time, indicating evolution.

Calculating Allele Frequencies

- It is calculated by dividing the number of times that particular allele appears by the total number of all alleles for that gene in the population.
- 1. Identify the allele:** Determine which specific allele's frequency you want to calculate.
 - 2. Count allele occurrences:** Count how many times that allele appears in the population.
 - 3. Count total allele copies:** Determine the total number of copies of all alleles at that gene locus in the population. For diploid, each individual has two copies of each gene.
 - 4. Calculate the frequency:** Divide the number of allele occurrences (step 2) by the total number of allele copies (step 3).

Equation, $p + q = 1$, which simply states that, if there are two alleles, their proportions must sum to 1

Let's say you're studying a gene with two alleles, **A** and **a**, in a population of 100 individuals. You count 120 A alleles and 80 a alleles.

- Total allele copies: 100 individuals * 2 alleles/individual = 200
- Frequency of A: $120 / 200 = 0.6$
- Frequency of a: $80 / 200 = 0.4$

So, the allele frequency of A is 0.6 (or 60%), and the allele frequency of a is 0.4 (or 40%)

$$P = \text{A Allele frequency} = 120/200 = 0.6$$

+

$$Q = \text{a Allele frequency} = 80/200 = 0.4$$

1

1

25 Mice

The following phenotype in mice :

BB=black, Bb= brown, bb= white

Calculate the allele frequency if there are 4 black, 8 brown and 13 white mice in gene pool ?

??

Calculate the allele frequency if there are 4 black, 8 brown and 13 white mice in gene pool ?

The following phenotype in mice :
BB=black, Bb= brown, bb= white

4= black BB

8 = brown Bb

13= white bb

25 mice = 50 Allele

$$B = (4 + 8) / 50 = 0.32$$

$$b = (13 + 8) / 50 = 0.68$$

Genotype frequencies

- Genotype frequencies refer to the proportions of different genotypes (combinations of alleles) within a population.
- Genotype frequency is calculated by dividing the number of individuals with a specific genotype by the total number of individuals in the population.

Calculating Genotype Frequencies

1. Identify the Genotypes

- For example, if you're looking at a gene with two alleles (let's call them A and a), the possible genotypes are AA, Aa, and aa.

2. Count Individuals

- Count how many individuals in the population have each genotype.

3. Calculate Frequencies

- For each genotype, divide the number of individuals with that genotype by the total number of individuals in the population.

Calculating Genotype Frequencies

- **Genotype frequencies describe the proportions of each genotype in the population.**

Example population data set:

- 98 pink-feathered birds with the AA genotype
 - 84 purple birds with the AB genotype
 - 18 blue birds with the BB genotype
= Total of 200 birds.
-
- The number of individuals with a genotype divided by the total number of individuals.
 - Frequency of the AA genotype is $98 \text{ divided by } 200 = 0.49$
 - Frequency of the AB genotype is $84 \text{ divided by } 200 = 0.42$
 - Frequency of the BB genotype is $18 \text{ divided by } 200 = 0.09$
 - The sum of these proportions is equal to 1.

Example

- Let's say you have a population of 100 fruit flies, and you are studying a gene for wing shape with two alleles: "W" for normal wings and "w" for vestigial wings. You observe the following:
- 64 flies have the genotype WW (normal wings)
- 32 flies have the genotype Ww (normal wings)
- 4 flies have the genotype ww (vestigial wings)

To calculate genotype frequencies:

- $f(WW) = 64 / 100 = 0.64$
- $f(Ww) = 32 / 100 = 0.32$
- $f(ww) = 4 / 100 = 0.04$

In a population in equilibrium there are 1000 people,
there are 160 recessive homozygous (aa) and
480 heterozygous genotypes (Aa).
Calculate the frequency of dominant allele ?

A & a allele

??

In a population in equilibrium there are 1000 people,
there are 160 recessive homozygous (aa) and
480 heterozygous genotypes (Aa).
Calculate the frequency of dominant allele ?

AA	Aa	aa
?	480	160 = 1000

360 +	480 +	160 = 1000
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$$A = 360 + 240 = 600/1000 = \mathbf{60\%}$$

$$a = 160 + 240 = 400/1000$$

Factors Influencing Genotype Frequencies

- Mutation → introduces new alleles
- Genetic drift → random changes
- Gene flow → mixing of populations
- Natural selection → changes fitness of genotypes
- Non-random mating

What is Genetic Drift?

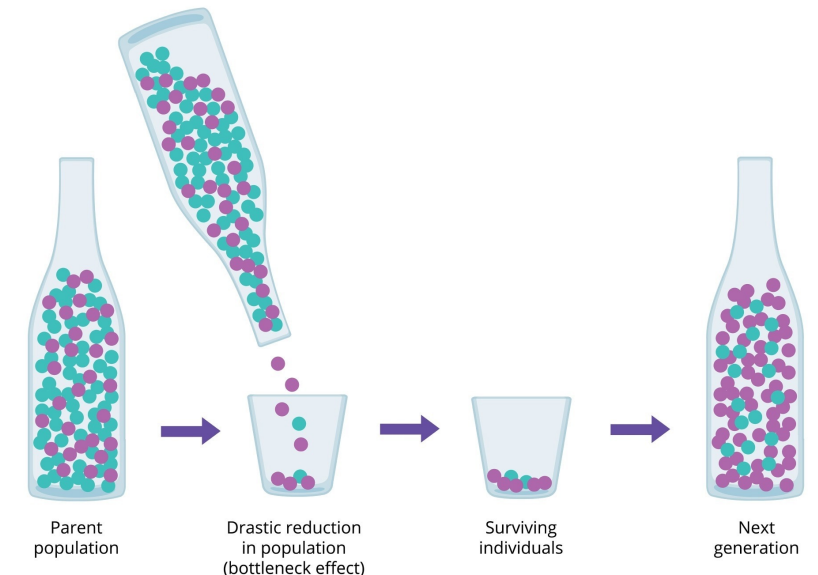
- Genetic drift is a mechanism of evolution in which allele frequencies within a population change randomly over time, due to chance events, rather than through natural selection.
- Genetic drift is change due to "sampling error" in selecting the alleles for the next generation from the gene pool of the current generation.
- Unlike natural selection (non-random), drift is **chance-driven**
- Stronger in **small populations**

Types of Genetic Drift

There are two main types of genetic drift

1. Bottleneck Effect

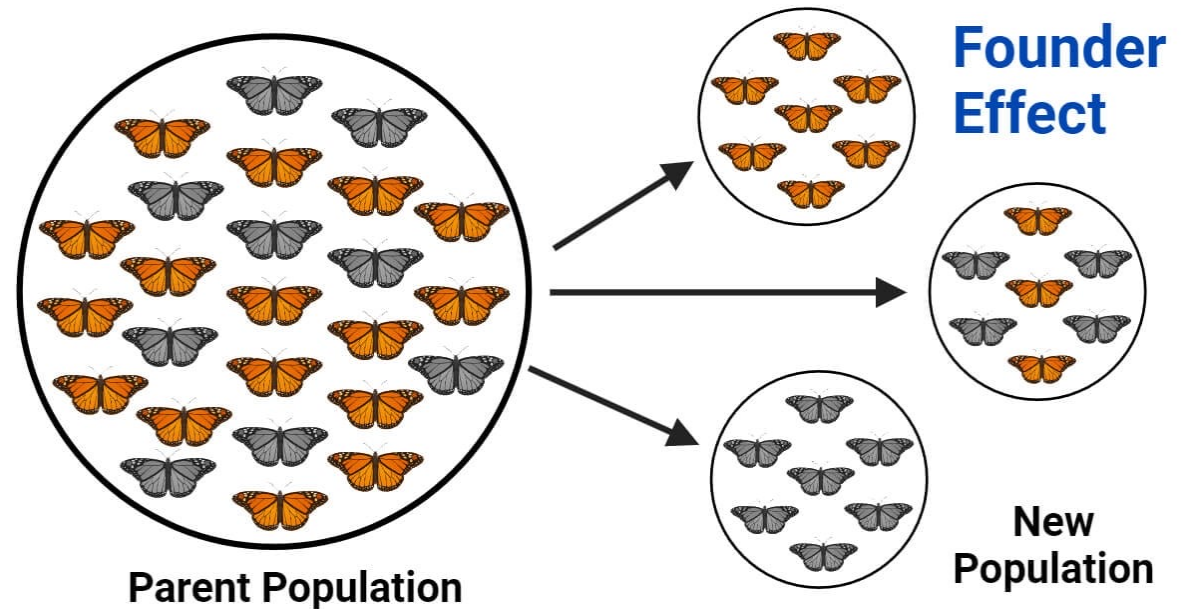
- This occurs when a population's size is drastically reduced due to a catastrophic event like a natural disaster (e.g., earthquake, flood, fire).
- The surviving individuals may not be representative of the original population's genetic diversity, leading to a loss of alleles and altered allele frequencies in the surviving population.
- The reduced genetic diversity makes the population more vulnerable to future environmental changes and diseases.



Types of Genetic Drift

2. Founder Effect

- This occurs when a small group of individuals from a larger population establishes a new, isolated population.
- The new population potentially leading to unique allele frequencies compared to the original, larger population.
- Over time, this small group, the "founders," can lead to significant differences in allele frequencies between the new and original populations.

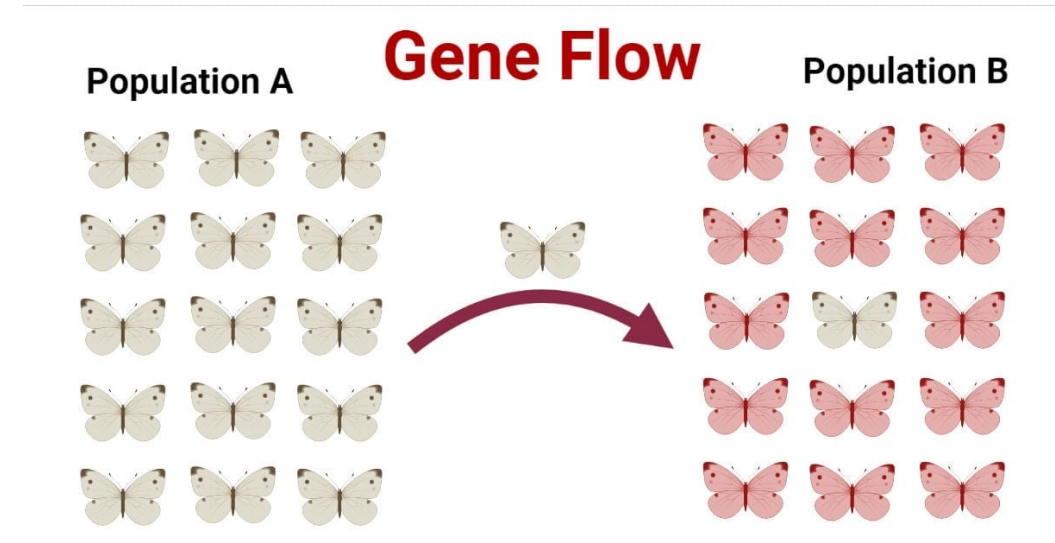


Consequences of Genetic Drift

- **Reduced genetic diversity:**
- Genetic drift can lead to the loss of alleles, especially in small populations, which can make them more vulnerable to environmental changes or diseases.
- **Fixation of alleles:**
- By chance, some alleles can become the only ones present in a population (fixed).
- **Can hinder adaptation:**
- Because genetic drift is random, it can lead to the loss of beneficial alleles or the fixation of less beneficial ones, potentially hindering a population's ability to adapt to its environment.

Gene Flow?

- Gene flow, also known as gene migration or allele flow, is the transfer of genetic material (genes or alleles) from one population to another, typically through interbreeding.
- Movement of alleles between populations
- Introduces new alleles into a population
- Introduces **new genetic variation**



Mechanisms of Gene Flow

- **Migration:** The movement of individuals from one population to another, carrying their genes with them, can be active (like animal migration) or passive (like seed dispersal).
- **Gamete transfer:** e.g., pollen dispersal in plants
- **Hybridization:** interbreeding between populations/species

Effects of Gene Flow

- Increases genetic diversity within populations
- Reduces genetic differences between populations
- Can spread beneficial alleles (adaptive introgression)
- Can also introduce maladaptive alleles

Natural selection

- Natural selection is a core concept in evolutionary biology where organisms with advantageous or favourable traits are more likely to survive and reproduce, passing those traits on to their offspring.
- Process where **alleles that improve fitness increase** in frequency

Types of Natural Selection

- **Directional selection** → One allele favored (shift in trait), For example, if taller giraffes are better at reaching food, directional selection would favor taller giraffes, causing the population's average height to increase over generations.
- **Stabilizing selection** → Intermediate favored (reduces extremes), For example, human birth weight tends to be within a certain range. Babies that are too small or too large may have lower survival rates, while those with average birth weights have a higher chance of survival.
- **Disruptive selection** → Favors both extreme traits, selecting against the intermediate ones. An example is a population of birds with beaks that can efficiently crack both very small and very large seeds. Birds with medium-sized beaks may be at a disadvantage when foraging for either type of seed.

Thanks