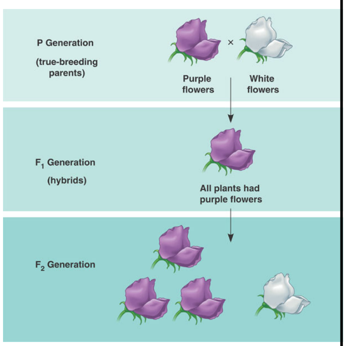
Basis of Population Genetics

Gregor Mendel

- Gregor Mendel was a monk who studied the garden pea (*Pisum sativum*).
  - Background in math & statistics
  - Emphasized experimentation
    - Replicates
  - Looked for patterns in his data

Gregor Mendel

- Mendel worked with several traits including flower color.
  - Started with true breeding parents.
  - All offspring were purple.
  - Next generation – white reappeared.
    - 3:1 ratio



Gregor Mendel

- Mendel discovered that characteristics pass from parent to offspring in form of discrete packets called **genes**.

  - Exist in alternate forms - **alleles**.
  - Some prevent expression of others.
    - **Dominant** vs. **recessive**
  - **Phenotype** – what the plant/fish looks like.
    - Purple or white
  - **Genotype** – which alleles are actually present.
    - PP, Pp or pp

Population Genetics

- Populations have genetic variation.
- Changes in the frequencies of alleles within a population is required for evolutionary change.
- Population genetics allows us to study how common a trait is in a population, and how that may change over time.

Population genetics

1. the translation of Darwin's three principles into genetic terms

a). Variation: Among individuals in a population there is phenotypic and genotypic variation

b). Heredity: Offspring are more similar to their parents than to unrelated individuals

c). Selection: Individuals having some phenotypes are more successful at surviving and reproducing than others

Population genetics

2. Study of heredity of traits controlled by one or a few genes in a population

a) description of genetic structure of a population (patterns of genetic variation found among individuals in a group)

b) examination of how genetic structure varies in space and time

c) evaluation of the processes that are responsible for producing genetic variation

Population genetics

3. Practical applications include importance for conservation biology and biodiversity

4. good example of the uses of mathematical theory in biology

Strongly dependent on mathematical models (which have been more successful than most areas of mathematical biology)

What is a population?

A population is a subdivision of a species

-A community of individuals where potential mates are usually found

- shares a common gene pool

- has continuity through time

- is linked by bonds or mating and parenthood

Population Genetics

•The genetic study of the process of evolution

•Deals with **frequencies of alleles & genotypes** in breeding populations.

It also deals with selective influences on the genetic composition of the population (*phenotypes*)

Aims

- To measure rates of change in the genetic composition of populations

- Helps to predict what alleles frequencies will be in the absence of evolutionary change.

Hardy Weinberg

- Hardy-Weinberg principle states that in a population mating at random **in the absence of evolutionary forces**, allele frequencies will remain constant from generation to generation.

$$p + q = 1$$
$$p^2+2pq+q^2 = 1.0$$

p = frequency of the most common allele
q = frequency of the less common allele

Genetic Structure of Populations

Genotype frequency

The proportion of a population that has one genotype relative to all genotypes at a species locus

$$f\text{ (particular genotype)} = \frac{\#\text{ individuals with that genotype}}{\text{total }\#\text{ individuals}}$$

Homozygotes: The two alleles are in the same state (AA, aa)

Heterozygotes: The two alleles are different (Aa)

Genetic Structure of Populations

Allele frequency

The proportion of one allele relative to all alleles at the locus in the population

f (particular allele) = # of copies of the allele in a population / sum of all alleles

i.e. Twice the number of homozygous genotypes with that allele (because homozygotes carry two copies each of the same allele) + the number of heterozygous genotypes with that allele (because heterozygotes carry only one copy of a particular allele), divided by two times the total number of individuals in the sample (because each individual carries two alleles per locus)

The Hardy-Weinberg principle (law)

Predicts the expected genotype frequencies using the allele frequencies in a diploid Mendelian population.

States that the frequencies of allele in a population will remain constant unless acted upon by outside agents of forces.

describes the genetics of **non-evolving populations**.

A non-evolving population is said to be in **Hardy-Weinberg Equilibrium**

In a population with diallelic locus (alleles A and B), if the frequency of one allele (A) is **p** and the other allele (B) is **q**, then.

Females	**Males** Freq (A) = p	Freq (B) = q
Freq (A) = p	Freq (AA) = pp	Freq (AB) = pq
Freq (B) = q	Freq (AB) = pq	Freq (B) = qq

Punnett square

If frequency (A) = p and frequency (a) = q,

Hence; p + q = 1 and 1- q = p

Then (p + q) = 1. This means $(p+q)^2 = 1$ too.
Therefore; $p(p+q)+q(p+q) = 1$
$p^2 + pq + pq + q^2 = 1$

$$p^2 + 2pq + q^2 = 1$$

WHERE; Frequency of the AA genotype = p^2
The frequency of the Aa genotype = 2pq
The frequency of the aa genotype = q^2

Hardy-Weinberg formula shows that: $p^2 + 2pq + q^2 = 1$

Since 'AA, Aa, aa' are the three possible genotypes for a biallelic locus, the sum of their frequencies should be 1.

HWE

$p^2+2pq+q^2 = 1$ *and* $p+q = 1$

p = frequency of the dominant allele in the population

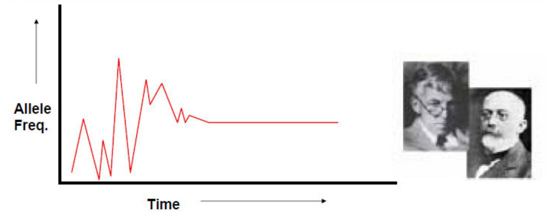
q = frequency of the recessive allele in the population

p^2 = percentage of homozygous dominant individuals

q^2 = percentage of homozygous recessive individuals

2pq = percentage of heterozygous individuals

Hardy-Weinberg Equilibrium



Example one locus (AA, Aa and aa)

If frequency of A gametes is 0.2. What is the proportion of the population that is the Aa genotype?

If frequency (A) = p and frequency (a) = q,
Hence; p + q = 1 :
thus 1 - 0.2 = q = ***0.8***

p^2	+ 2pq	+ q^2	=	1
0.2(0.2)	+ 2pq	+ 0.8(0.8)	=	1
0.04	+ 2pq	+ 0.64	=	1

2pq = 1 – 0.68 = <u>0.32</u> or
2pq = 2(0.2X0.8) = <u>0.32</u>

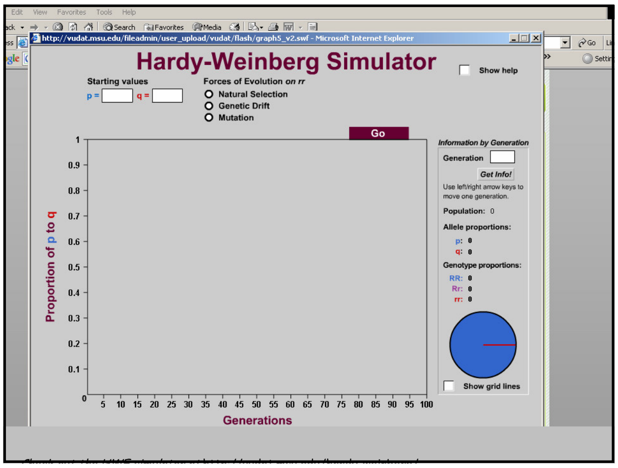
Calculating the gene frequency (two ways)

Suppose that we have 200 individuals: 83 AA, 62 AB, 55 BB

Method 2. Calculate what fraction of genes/alleles in the parents that are **A**:

Genotype	Number	A's	B's
AA	83	166	0
AB	62	62	62
BB	55	0	110
		228	172

Answer. 228/400 = **<u>0.57 *A*</u>**



Genetics - the field of science that studies genes & their:
1) diversity of forms
2) mutations
3) replication, and
4) translation of information.

Chapter 1 Opener
Introduction to Genetic Analysis, Ninth Edition
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Genetic variation in the color of corn kernels

Gene – the basic unit of inheritance

Trait – an inherited characteristic
AKA- *phenotype*

Genetic variation within a population

A. <u>Polymorphism</u> = genetic variation; the occurrence of several phenotypic forms of a character associated with one locus (gene) or homologues of one chromosome

Types of polymorphisms that population geneticists examine;

1. Morphological polymorphisms
2. Chromosomal polymorphisms
3. Immunological polymorphisms
4. Protein polymorphisms
5. DNA sequence polymorphisms

B. <u>Heterozygosity</u> = measure of the frequency of the heterozygote genotype at a loci or at multiple loci

What makes a population evolve or disrupt the HWE equilibrium?

- **mutation**
- migration
- natural selection
- genetic drift
- non-random mating

spontaneous change in DNA

- creates new alleles
- ultimate source of all genetic variation

more mutations per generation	→ higher frequency of mutant allele at equilibrium
stronger selection against mutant allele	→ lower frequency of mutant allele at equilibrium

What can change population genetic structure?

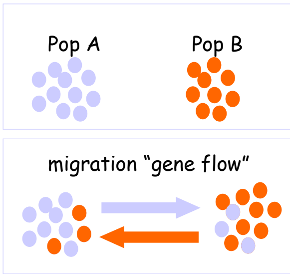
- mutation /recombination
- migration
- natural selection
- genetic drift
- non-random mating

creating new combinations of existing diversity

New genes and new alleles originate only by mutations can change allele frequency

What can change population genetic structure?

- mutation
- **migration/gene flow**
- natural selection
- genetic drift
- non-random mating



Will significantly change allele frequency if some alleles tend to migrate more often than others

individuals move into populations

- *introduces new alleles*

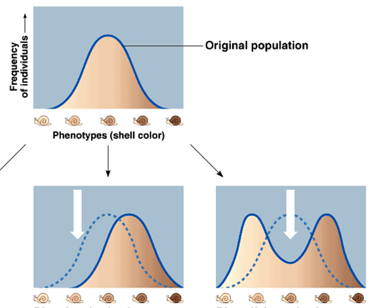
What can change population genetic structure?

- mutation
- migration
- **natural selection**
- genetic drift
- non-random mating

Certain genotypes produce more offspring

- differences in survival or reproduction
- leads to adaptation

Modes of selection

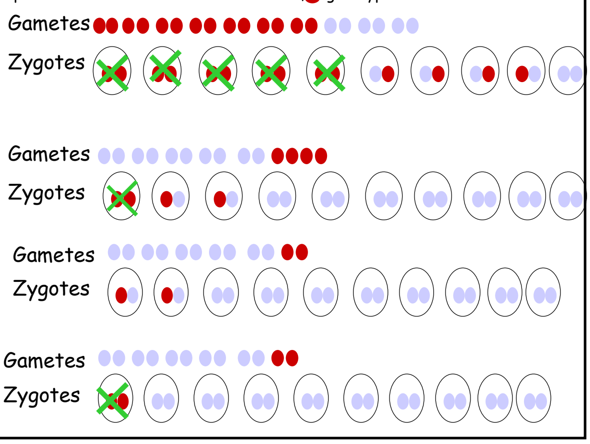


Stabilizing selection culls extreme variants from the population, in this case eliminating individuals that are unusually light or dark. The trend is toward reduced phenotypic variation and maintenance of the status quo.

Directional selection shifts the overall makeup of the population by favoring variants of one extreme. In this case, the trend is toward darker color, perhaps because the landscape has been blackened by lava.

Diversifying selection favors variants of opposite extremes over intermediate individuals. Here, the relative frequen-cies of very light and very dark snails have increased. Perhaps the snails have colonized a patchy habitat where a background of white sand is studded with lava rocks.

The process of natural selection at one locus, genotypes are lethal in this case



Natural selection

Some alleles will be preferentially represented in the next generation

- Traits that result in differential success in reproduction

- Includes traits that increase survival an individual that lives longer will reproduce more than a shorter lived one

- And traits that help the individual reproduce more often sexual selection
    - Leads to adaptive variation
    - Genetic variation is the raw material for natural selection

- Natural selection will act upon the variation that is already there

What can change population genetic structure?

- mutation
- migration
- natural selection
- **genetic drift**
- non-random mating

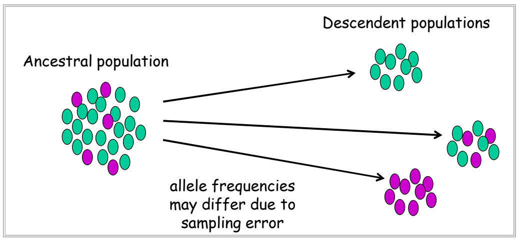
genetic change by chance alone

- sampling error
- misrepresentation
- small populations

Forms of genetic drift

1. Founder effect

- a small group leaves a large population and starts a new population
- only some alleles are sampled from the gene pool

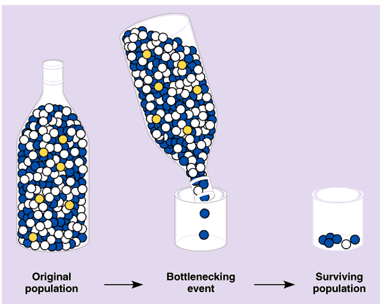


2. Population bottleneck

– large population shrinks to a small number of individuals, which reproduce to repopulate

allele frequencies may differ due to sampling error

In this case
Yellow allele is lost



What can change population genetic structure?

- mutation
- migration
- natural selection
- genetic drift
- **non-random mating**

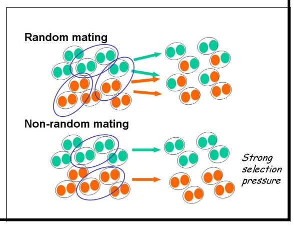
- Inbreeding
- Assortative mating

mating combines alleles into genotypes

- non-random mating

⇩

non-random
allele combinations



Of the five conditions for H-W equilibrium, which ones change allele frequency, and which ones change phenotypic frequency?

• Mutation (negligible in most pop) • Natural selection • Migration • Genetic drift	Allele freq
• Non-random mating	Phenotype freq

A population won't evolve if there is…

- an infinitely large population (no genetic drift)
- no change from one allele to the other (no mutation)
- Equality in genotype viability (no natural selection
- no migration (no gene flow)
- Equality in mate choice (random mating)

…then, the population is said to be in Hardy-Weinberg equilibrium

NOTE!

- The Hardy-Weinberg law rarely holds true in nature (otherwise evolution would not occur).
- Organisms are subject to mutations, selective forces and they move about, or the allele frequencies may be different in males and females.
- Gene frequencies are constantly changing in a population, but the effects of these processes can be assessed by using the Hardy-Weinberg law as the starting point.
- **HWE** is the null hypothesis of evolution

Population Structure

A population is considered structured if:

- genetic drift is occurring in some of its subpopulations

- migration does not happen uniformly throughout the population, or

- mating is not random throughout the population.

A population's structure affects the extent of genetic variation and its patterns of distribution.

Factors causing genotype frequency changes or evolutionary principles

- **Selection** = variation in fitness; heritable
- **Mutation** = change in DNA of genes
- **Migration** = movement of genes across populations
- **Recombination** = exchange of gene segments
- **Non-random Mating** = mating between neighbors rather than by chance
- **Random Genetic Drift** = if populations are small enough, by chance, sampling will result in a different allele frequency from one generation to the next

Assumptions (HWE)

1. The **absence of evolutionary process** (e.g., mutation, migration, drift, selection) affecting the allele frequencies in the population

2. **Random mating**- the probability of mating between individuals is independent of their genetic constitution—no assortative mating

3. **Large population size** i.e., population size is effectively infinite. To reduce sampling error with generations

4. Males and females have **similar allele frequencies**.

5. **There is no selection**. Lack of differential survival and reproductive success i.e. All genotypes reproduce with success.

6. Organism is **diploid**

Hardy-Weinberg equilibrium

assumptions:

how the model works — diploid, sexual

which alleles are passed on — no selection, no mutation, no migration, no genetic drift

how alleles are combined into genotypes — random mating

IF: these assumptions are true

THEN: no change in allele or genotype freq's

Forces of evolution

How the forces of evolution Increase (+) or decrease variation between populations

Force	Variation within populations	Variation between populations
Inbreeding or genetic drift	–	+
Mutation	+	–
Migration	+	–
Selection		
Directional	–	+/–
Balancing	+	–