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Paper 401: Animal Taxonomy and Evolution

Topic: Hardy-Weinberg law of equilibrium and destabilizing forces

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M.Sc. - ZOOLOGY IV SEMESTER

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INTRODUCTION

- The rediscovery of Mendel's work in the 1900 began a 30-year effort to reconcile Mendel's concept of genes and alleles with the theory of evolution.
- Alfred R. Wallace and Charles Darwin first identified natural selection as the mechanism of evolution in the mid 19th century but had no accurate model to depict the mechanism responsible for this process.
- Biologists later realized that the phenotypic traits in a population is linked to the relative abundance of the alleles influencing those traits.
- Population genetics then came into the spotlight which started the study of genetic variation in a population and how it changes over time.
- Experiments are done to collect data that represent the allele frequencies in a normal population and those of in a population acted upon by the evolutionary forces.

POPULATION

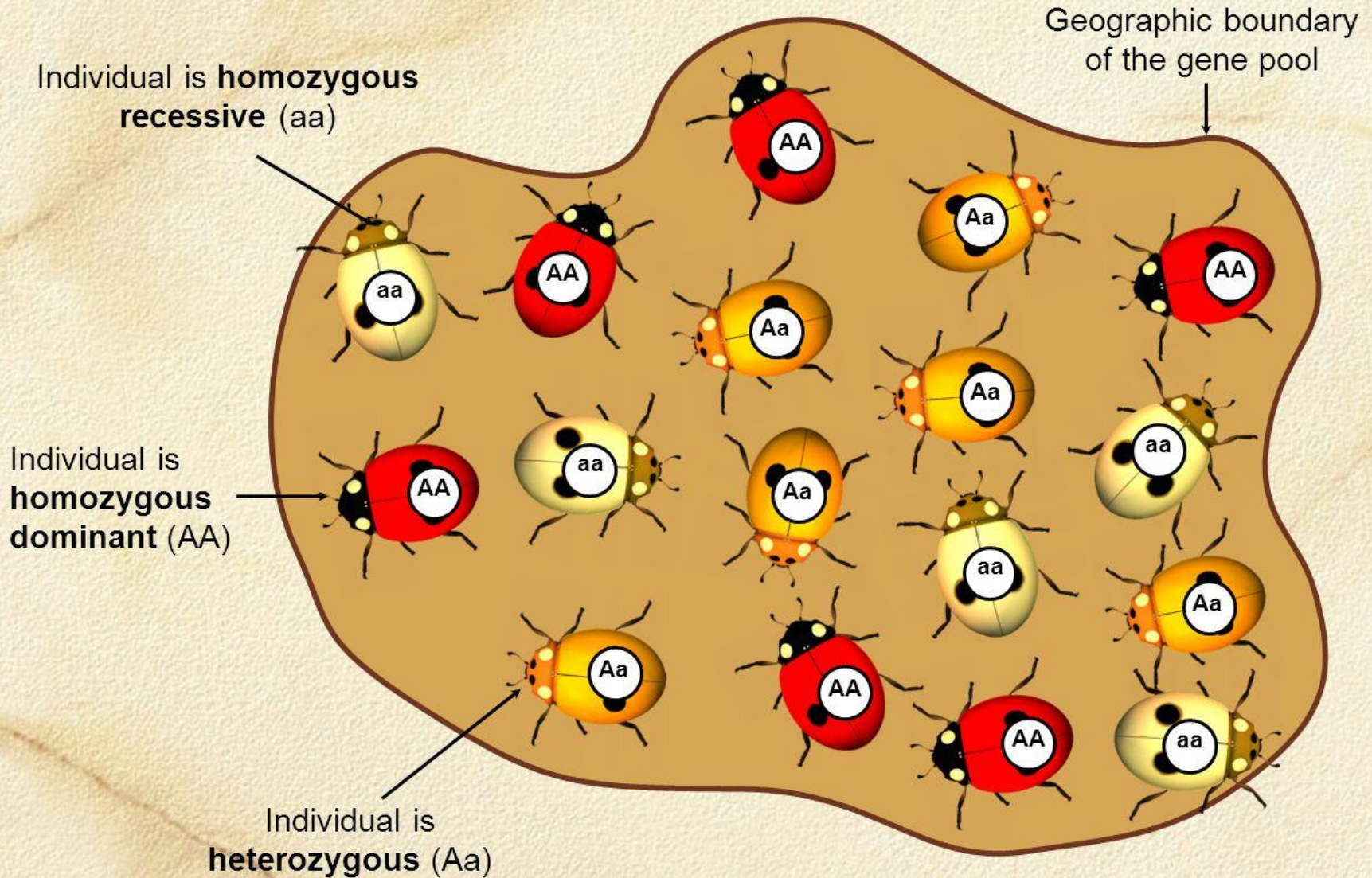
- A population is a group of sexually interbreeding individuals present in a given geographical area.
- Transmission of genes occur in accordance with Mendelian laws.
- A population has somewhat similar genetic constitution and gene alignment except for some uniqueness.
- It has a given gene pool which is shared by all of them.
- Free gene flow among the individuals takes place.
- Each member has equal chance of mating with any other member of opposite sex.
- Gene reshuffling takes place.



GENE POOL

- ❖ It is the sum total of genes present in a given population.
- ❖ These genes are transferred to the next generation.
- ❖ Study of gene pool of a population tells us about
 - Total number of genes
 - Kinds and varieties of genes
 - Proportion
 - Distribution
- ❖ Maintains its integrity as long as there is no interbreeding.
- ❖ Tries to maintain its genetic equilibrium.
- ❖ Size of a gene pool depends on the number of genes and the number of individuals carrying these genes.

Gene Pool



A gene pool made up of 16 individual organisms with gene A, and where gene A has two alleles

GENE FREQUENCY

- It refers to the proportion of an allele in the gene pool of a population as compared with other genes at the same locus.
- It can be calculated by dividing the number of a particular allele in question with the total number of alleles present on that locus in the population.
- For a gene with 2 alleles say A and a has the following proportion of contribution.
- In a population
 - N = total number of individuals in the population.
 - D = number of homozygous dominants
 - H = number of heterozygous individuals
 - R = number of homozygous recessives
- Calculating gene frequency in a given population :
 - Total number of individuals = N
 - Total number of alleles = $2N$
 - No. of homozygous dominants = D
 - No. of recessive homozygous = R
 - No. of hetero zygotes = H
 - Total no. of dominant alleles = $2D+H$
 - Total no. of recessive alleles = $2R+H$
 - Frequency of dominant gene 'A' = $2D+H/2N$
 - Frequency of recessive allele 'a' = $2R+H/2N$

GENOTYPE FREQUENCY

- It is the total number of one kind of individuals from a population all of which exhibit similar character with respect to the locus under consideration.
- It can be calculated by dividing the number of individuals with that genotype by the total number of individuals in the population.
- Naturally, Three kinds of individuals occur in the population :
 - Homozygous dominant
 - Heterozygous and
 - Homozygous recessive
- Calculation of genotype frequency :
 - Allele frequency of 'A' allele = p
 - Allele frequency of 'a' allele = q
 - Genotype frequency of AA = p^2
 - Genotype frequency of aa = q^2
 - Genotype frequency of Aa = $2pq$

HARDY-WEINBERG'S LAW OF EQUILIBRIUM

- The fundamental idea in calculating and generating the idea of distribution of a gene in a population was offered by English man, G.H Hardy and German man, W. Weinberg simultaneously in the year 1908.
- It lead to the foundation of population genetics and of modern evolutionary theory in the form of what we now call as Hardy-Weinberg's law of equilibrium.
- According to this law,“The relative frequencies of various kinds of alleles in a large and randomly mating panmictic population tend to remain constant from generation in the absence of certain destabilizing forces.”
- The relationship between gene frequency and genotype frequency can be expressed as : $(p+q)^2= 1$

where, p is the frequency of allele 'A'

q is the frequency of allele 'a'

The Equations

$$p + q = 1$$

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

- A gene has two alleles, A and a
- The frequency of allele A is represented by p
- The frequency of allele a is represented by q
- The frequency of genotype $AA = p^2$
- The frequency of genotype $aa = q^2$
- The frequency of genotype $Aa = 2pq$

SALIENT FEATURES OF HARDY-WEINBERG'S LAW

- ❑ According to Hardy-Weinberg law, the gene and genotype frequencies of each allele in a population remains at equilibrium generation after generation, if it exhibits following attributes:
 - Random mating
 - Large population size
 - Bi-parental mode of reproduction
 - Homogenous age structure
 - Absence of evolutionary forces
 - No preferential mating

EXAMPLES

Frequencies of multiple alleles

- For ABO blood group:

Blood type	Genotype	Frequency	Total
A	$I^A I^A$	p^2	$p^2 + 2pr$
	$I^A i$	$2pr$	
B	$I^B I^B$	q^2	$q^2 + 2qr$
	$I^B i$	$2qr$	
AB	$I^A I^B$	$2pq$	$2pq$
O	ii	r^2	r^2

In a pygmy group in Central Africa, the frequencies of alleles determining the ABO blood groups were estimated as 0.74 for I^O , 0.16 for I^A , and 0.10 for I^B . Assuming random mating, what are the expected frequencies of ABO genotypes and phenotypes?

Genotype	Phenotype	Frequency
$I^O I^O$	Type O	$p^2 = 0.74 * 0.74 = \mathbf{0.548}$
$I^A I^O$	Type A	$2pq = 2 * 0.74 * 0.16 = \mathbf{0.237}$
$I^A I^A$	Type A	$q^2 = 0.16 * 0.16 = \mathbf{0.026}$
$I^B I^O$	Type B	$2pr = 2 * 0.74 * 0.10 = \mathbf{0.148}$
$I^B I^B$	Type B	$r^2 = 0.10 * 0.10 = \mathbf{0.01}$
$I^A I^B$	Type AB	$2qr = 2 * 0.16 * 0.10 = \mathbf{0.032}$

Hardy-Weinberg for X-linked alleles:

e.g., Humans and *Drosophila* (XX = female, XY = male)

	X ^A (p)	X ^a (q)	Y
X ^A (p)	X ^A X ^A p ²	X ^A X ^a pq	X ^A Y p
X ^a (q)	X ^A X ^a qp	X ^a X ^a q ²	X ^a Y q

Females

- Hardy-Weinberg frequencies are the same for any other locus:
 $p^2 + 2pq + q^2 = 1$

Males

- Genotype frequencies are the same as allele frequencies:
 $p + q = 1$
- Recessive X-linked traits are more common among males.

DESTABLIZING FORCES OF HARDY-WEINBERG'S LAW

- A natural biological population is dynamic which is at par as per the theoretical static Hardy-Weinberg model of genetic population.
- Its gene equilibrium changes from one generation to the next generation by the disruptive action of evolutionary forces.
- Study of the effect of certain destabilizing forces was done by R.A. Fischer, Sewall Wright and J.B.S. Haldane and thus the dynamics of changing population was emphasized.
- The evolutionary forces are those that change the gene pool of the population and thus evolution takes place.

DESTABILIZING FORCES

Natural selection

Mutation

Genetic drift

Migration

Meiotic drive

NATURAL SELECTION

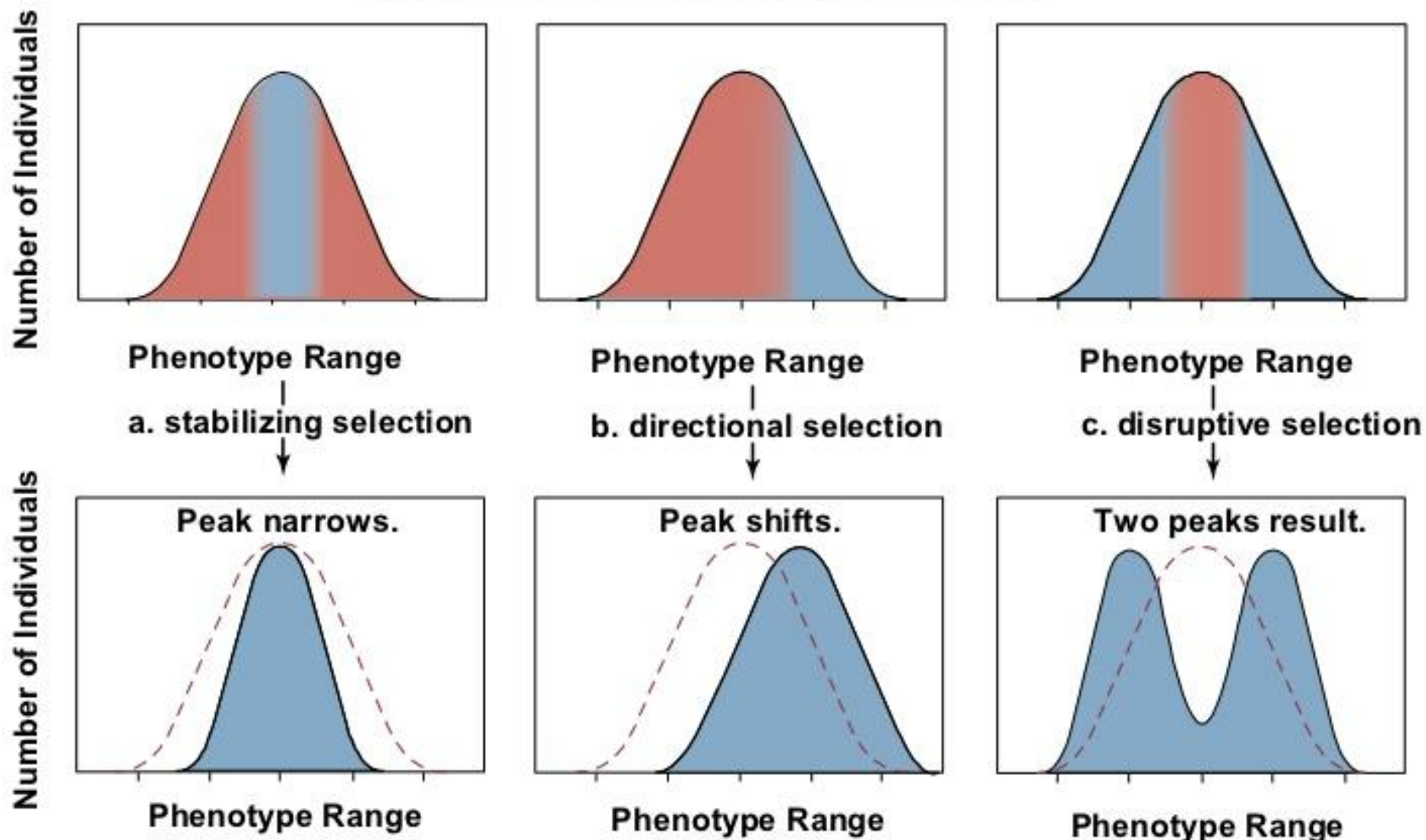
- Natural selection as the guiding force of evolution was recognised by Charles Darwin. But his concept was applicable to individuals rather than to the populations.
- It may be defined as the force which maintains consistent differences in the contributions of various genotypes to the next generation.
- According to Dobzhansky, it is the differential contribution of the alleles of a gene to the gene pool of a population in the next generation.
- It is the genotype which actually changes under the influence of environment, variations caused by changes in the genes and in the chromosomes which produce heritable variations in the organism and thus in the population.

TYPES OF NATURAL SELECTION

- ❑ Based on the different organisms and environment relationship, following different kinds of natural selection have been recognised:
 - Stabilising
 - Directional
 - Disruptive
 - Cyclic

Figure 13.15A Phenotype ranges before and after three types of selection
Blue represents favored phenotype(s)

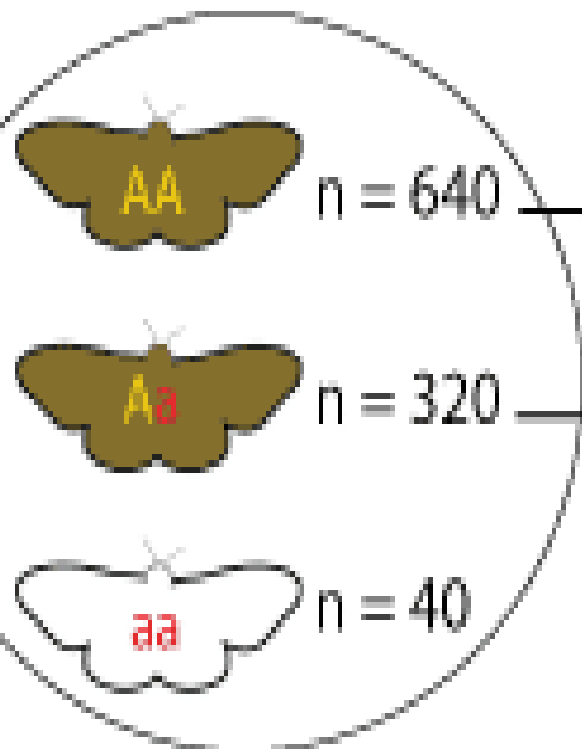
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Initial population

Selection

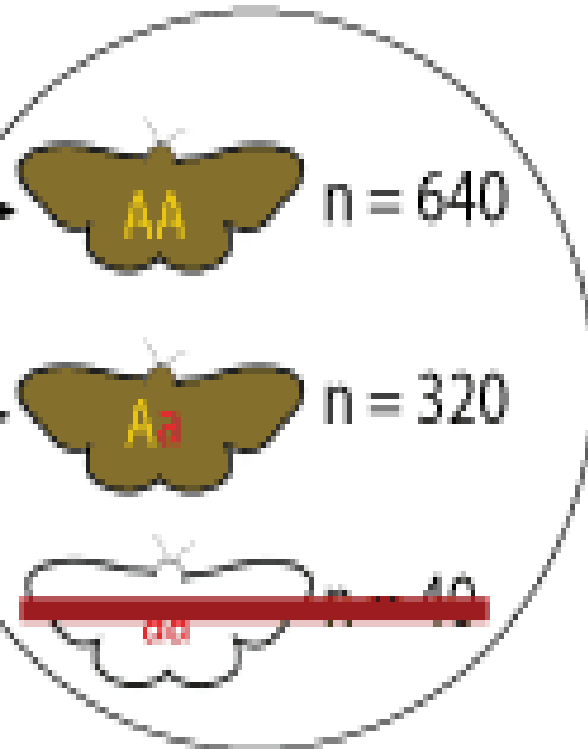
Next generation



survives

survives

X



Population (n=1000)
 $p = 0.8, q = 0.2$

Population (n=960)
Total No. alleles: $960 \times 2 = 1920$
 $p = (640 + 640 + 320) / 1920 = 0.833$
 $q = 320 / 1920 = 0.167$

MUTATION

- ❖ It can be stated as any change in the normal nucleotide sequences of a gene.
- ❖ It acts to create new alleles that changes the normal equilibrated gene pool of a population.
- ❖ It occurs at random i.e. without any regard for any possible benefit or disadvantage to the organism.
- ❖ To see the effect of any mutation that occurred in a population :
 - The allele evaluated must produce a distinctive phenotype that can be distinguished from a normal phenotype.
 - The trait must be fully expressed.
 - An identical phenotype must never be produced by a non genetic agents such as drugs or chemical.

Change in allele frequency due to mutation

$$\Delta q = \mu p - \nu q$$

Where,

μ = rate of forward mutation (From A1 to A2)

ν = rate of backward mutation (From A2 to A1)

At equilibrium, $\Delta q=0$,

$$q_{eq} = \frac{\mu}{\mu + \nu}$$

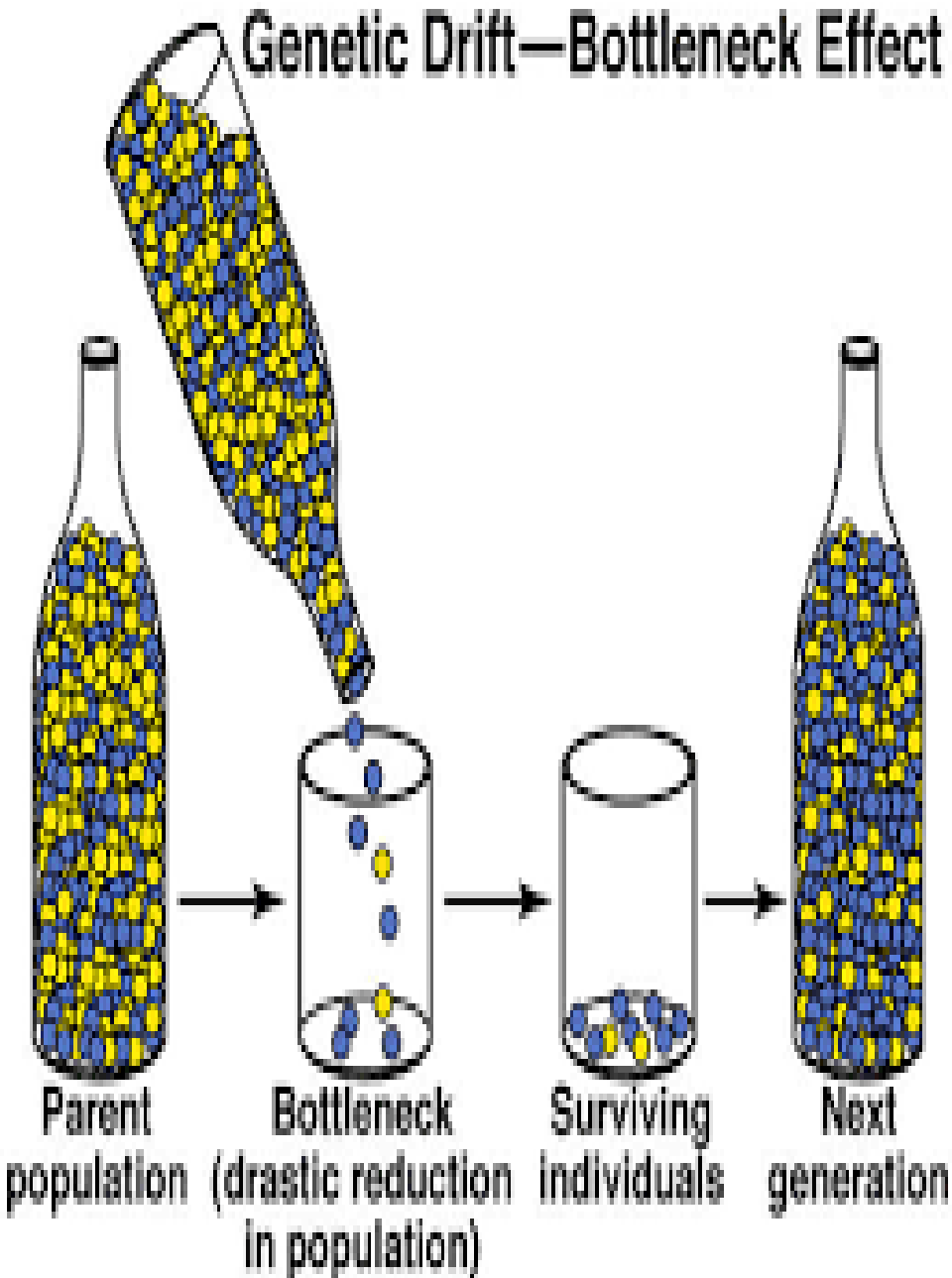
Assumptions:

1. Large population size.
2. No selection.

GENETIC DRIFT

- In small populations, significant random fluctuations in allele frequencies are possible through chance deviation.
- The degree of fluctuation increases as the population size decreases.
- This fluctuation due to chance is known as the **genetic drift**.
- Genetic drift can arise due to :
 - **Founder effect** : The genetic consequences seen when a population originates from a small number of individuals.
 - **Bottleneck effect**: It develops when a large population undergoes a drastic but temporary reduction in numbers.
- It occurs when the number of reproducing individuals in a population is too small to ensure that all the alleles in the gene pool will be passed on to the next generation in their existing frequencies.

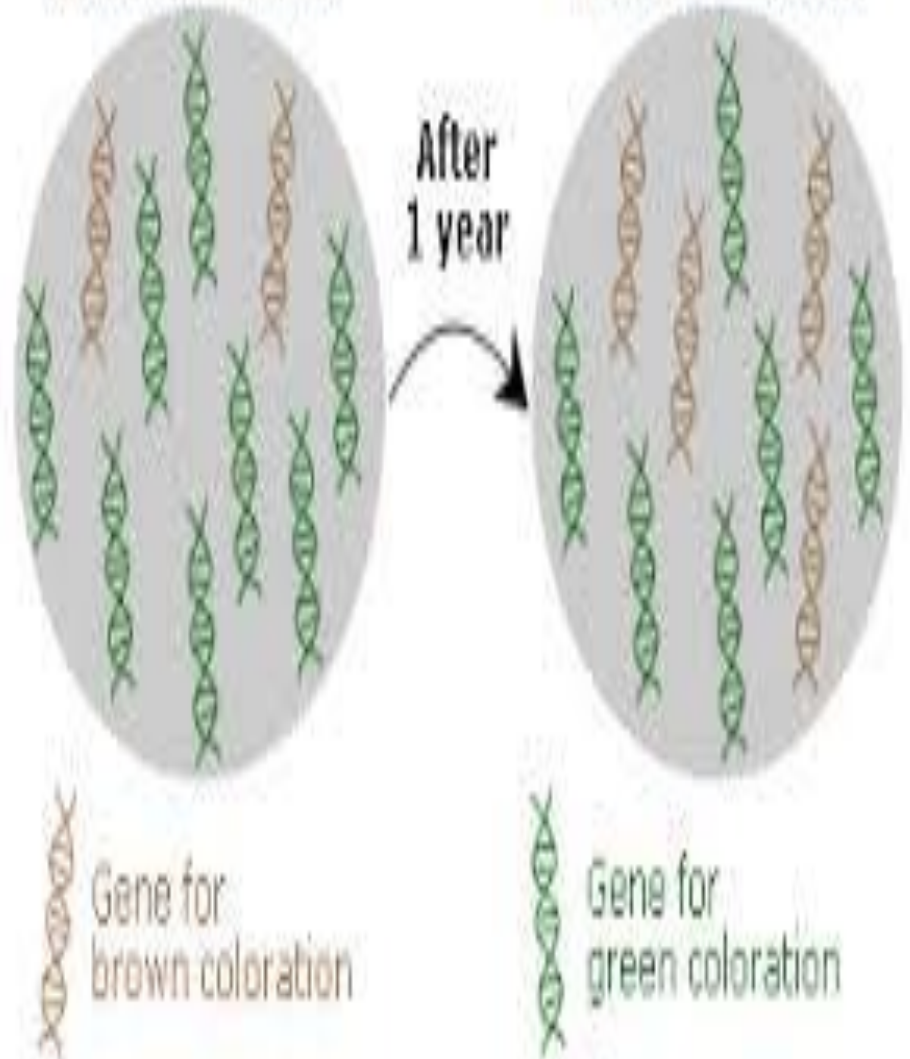
Genetic Drift—Bottleneck Effect



Bottleneck Effect

80% Green Genes
20% Brown Genes

60% Green Genes
40% Brown Genes



Founder Effect

NONRANDOM MATING

- ❖ It changes the frequencies of genotypes in a given population.
- ❖ Any one genotype has an equal probability of mating with any other genotype in the population.
- ❖ Subsequently, selection for or against certain genotypes has the potential to affect the overall frequencies of the alleles they contain.
- ❖ Non random mating can take one of several forms:
 - **Positive assortative mating** : Similar genotypes are more likely to mate than dissimilar ones.
 - **Negative assortative mating** : Dissimilar genotypes are more likely to mate.

How does **non-random mating** affect pop'n genetic structure?

Example of assortative mating

Generation 1

25% AA
50% Aa
25% aa

all AA mate with AA
all Aa mate with Aa
all aa mate with aa

{ 25% AA }
{ 12.5% AA }
25% Aa
{ 12.5% aa }
{ 25% aa }

Generation 2

37.5% AA
25% Aa
37.5% aa

Allele freq:
0.5 A 0.5 a

Did allele frequencies change?

Allele freq:
0.5 A 0.5 a

*Is the population in
Hardy-Weinberg equilibrium?*

Assortative mating leads to excess homozygotes

MIGRATION

- ❖ A species become divided into populations that are separated geographically.
- ❖ Migration occurs when individuals move between the populations.
- ❖ For example :
 - In a given species a given locus has 2 alleles, A and a.
 - There are two populations of this species, one on the mainland and one on an island.
 - The frequency of A on the mainland is represented by p_m and the frequency of a on the island is p_i .
 - If there occurs migration from the mainland to the island , the frequency of A in the next generation on the island is given by

$$p_i = (1-m) p_i + m p_m$$

where m represents migrants from the mainland to the island.

Changes in allele frequency due to migration

m_i = gene flow = $\frac{\# \text{ breeding immigrants from donor population } j}{\text{size of recipient population } i}$

migrants (m) moving from donor (j) to recipient (i)

Change in allele frequency (q) in population i:

	Before	After
Recipient i	q_i	$q_i' = (1 - m_j) q_i + m_j q_j$
Donor j	q_j	q_j



SIGNIFICANCE OF HARDY WEINBERG'S LAW

- Describes a situation in which there is genetic equilibrium and no evolution.
- Provides a theoretical base for measuring evolutionary change.
- Conserves gains made in the past.
- Avoids too rapid changes.
- Maintains heterozygosity in the population.
- Prevents evolutionary progress.

CONCLUSION

- No population is ideal as per the Mendelian laws. It undergoes dynamic changes that alter the gene frequencies which ultimately brings about a new and different gene pool for a population.
- These destabilizing forces add on or remove genes that changes the allele frequencies that causes a new species or a new population to be formed.

REFERENCES

- Strikberger, M.W.Jones and Bartett Publisher, Boston London.
- T. Strachan & A.P Read, Human Molecular Genetics, Blackwell.
- A textbook on Organic Evolution by V. B. Rastogi.
- www.biologydiscussion.com
- www.sciencedirect.com