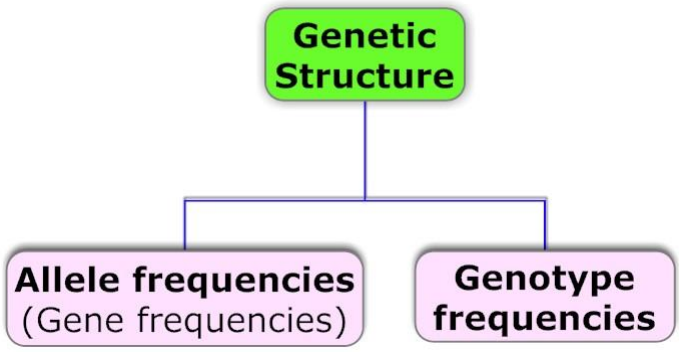
POPULATION GENETICS

- A population is a set of organisms in which any pair of members can breed together. I t may be a local population such as the cattle in a farm or it could be as large as an entire species.
- Population genetics is concerned with the genetic constitution (allele and genotype frequencies) of populations, their relationship and how this constitution changes with time.
- Population genetics describes how genetic transmission happens between a population of parents and a population of offspring.
- Traditionally, the study of population genetics involved the identification of different alleles through observation of the expressed traits, broadly called the phenotype.
- Alleles are different versions of the same gene that are expressed as different phenotypes.
- Every diploid individual has two copies (two alleles) of each gene, one inherited from each parent.
    - If an individual has two different versions of a particular gene, the individual is said to be heterozygous for that gene.
    - If the two alleles are the same, the individual is homozygous for that gene.
- In the transmission, the genotypes of the parents are broken down and a new set of genotypes is constituted in the progeny, from the genes transmitted in the gametes.
- The genes carried by the population thus have continuity from generation to generation, but the genotypes in which they appear do not.
- *Gene pool* is defined as the sum total of genes present in a Mendelian population. It includes all the genes of all the individuals of a population.
- The transfer of genes from one gene pool to another is called *gene flow*.
- The classical Mendelian traits which are qualitative in nature can be classified into distinct phenotypic classes/categories.
- These traits are controlled by only one or a very few genes with almost no environmental effect to modify the gene effects.
- Population genetics was first used as the basis for genetic improvement of livestock by J.L.Lush

GENETIC STRUCTURE OF POPULATIONS



GENE FREQUENCY

- Standard usage in population genetics uses the term gene frequency for what is actually allele frequency.
- Gene or allele frequency is the proportion of one allele relative to all alleles at the locus in the population.

$$f(allele) = \frac{No.of\ copies\ of\ the\ allele\ in\ a\ population}{Sum\ of\ all\ alleles}$$

- Gene frequency is usually expressed as a proportion or a percentage.
- The frequency of a phenotype in a population depends on the frequency of the allele controlling it.
- Frequently used over genotypic frequencies in calculations because
  - Genotypes break down to alleles when gametes are formed
  - Alleles (not genotypes) are passed on to progeny
- If *f(AA)*, *f(Aa)*, and *f(aa)* are the frequencies of the three genotypes at a locus with two alleles "*A*" and "*a*"

The frequency of the *A*-allele

- $f(A) = p$

The frequency of the *a*-allele

- $f(a) = q$

$$f(A) + f(a) = p + q = 1$$

- Calculation from observed number of individuals with each genotype

$$f(A) = p = f(AA) + \frac{1}{2}f(Aa)$$

$$f(a) = q = f(aa) + \frac{1}{2}f(Aa)$$

$$p + q = f(AA) + f(Aa) + f(aa) = 1$$

from this we get

$$q = 1 - p \quad and \quad p = 1 - q$$

GENOTYPE FREQUENCY

- Genotype frequency is the proportion of a population that has one genotype relative to all genotypes at a specific locus.
- For example: Two alleles are possible - *A* or *a*, that can combine to give the following genotypes *AA* (homozygous dominant), *Aa* (heterozygous), *aa* (homozygous recessive).

Genotype	No. of individuals	Genotype frequency
AA	60	f(AA) = 60/200 = 0.3 or 30%
Aa	100	f(Aa) = 100/200 = 0.5 or 50%
aa	40	f(aa) = 40/200 = 0.2 or 20%
Total	**200**	=1.0 or 100%

GENETIC VARIATION

- Some of the causes for genetic variation being present within a population are
  - Polymorphism
  - Crossing over
  - Independent assortment of the homologous pairs of chromatids
  - Mutation - gene and chromosome
  - Random fertilisation

FACTORS AFFECTING GENETIC CONSTITUTION

Factors affecting genetic constitution of a population

- Selection
  - Variation in fitness and heritability favors a single phenotype and therefore allele frequency continuously shifts in one direction
- Mutation

- Change in DNA of genes and thereby introduce new allele are introduced into the gene pool

- Migration
  - Migration means movement of individuals
  - This movement introduces new alleles into the population (gene flow).
  - The gene frequencies in the population may be changed by immigration of individuals from another population or emigration of individuals from the population.
  - Dispersal of animals, pollen on the wind etc.
- Recombination
  - Exchange of gene segments shuffle the genes into new combinations which can result in organisms exhibiting different traits.
- Mating system
  - Mating system two types: Random mating (Panmixia) and Non-random mating.
  - Individuals those are more closely (inbreeding) or less closely related mate more often rather than by chance.
  - Resulting in c hange of genetic constitution of a population.
- Random Genetic Drift
  - If populations are small enough, by chance, sampling will result in a different allele frequency from one generation to the next.
- Differences in fertility and viability
  - The individuals in a population (different genotypes) differ in fertility and contribute unequally to the next generation thus changing the gene frequency in the subsequent generation.

MODULE-22: HARDY WEINBERG LAW

Learning objectives

After completing this module, the learner should be able to:

- understand the concept of Hardy - Weinberg Law
- understand the five conditions that hold for Hardy-Weinberg Equilibrium
- predict genotype frequencies in a population given allele frequencies and assuming random mating using Hardy - Weinberg Proportions

HARDY- WEINBERG LAW

- Hardy-Weinberg law was proposed independently in 1908 by Wilhelm Weinberg, a German physician, and Godfrey Harold Hardy, a British mathematician.

Both gene and genotype frequencies in a population remain constant generation after generation when the population is large; mating is at random and in the absence of selection, mutation and migration.

- When the gene frequency remains constant generations after generations, the population is in genetic equilibrium or Hardy-Weinberg equilibrium non-evolutionary model.
- When the population is in genetic equilibrium, the rate of evolution is zero. That is, when a population obeys, hardy-Weinberg law the population will not undergo evolution. So evolution occurs only when Hardy-Weinberg equilibrium is altered.
- The Hardy-Weinberg law is represented by a simple formula.
    - For 2 alleles (A_1 and A_2) of one gene

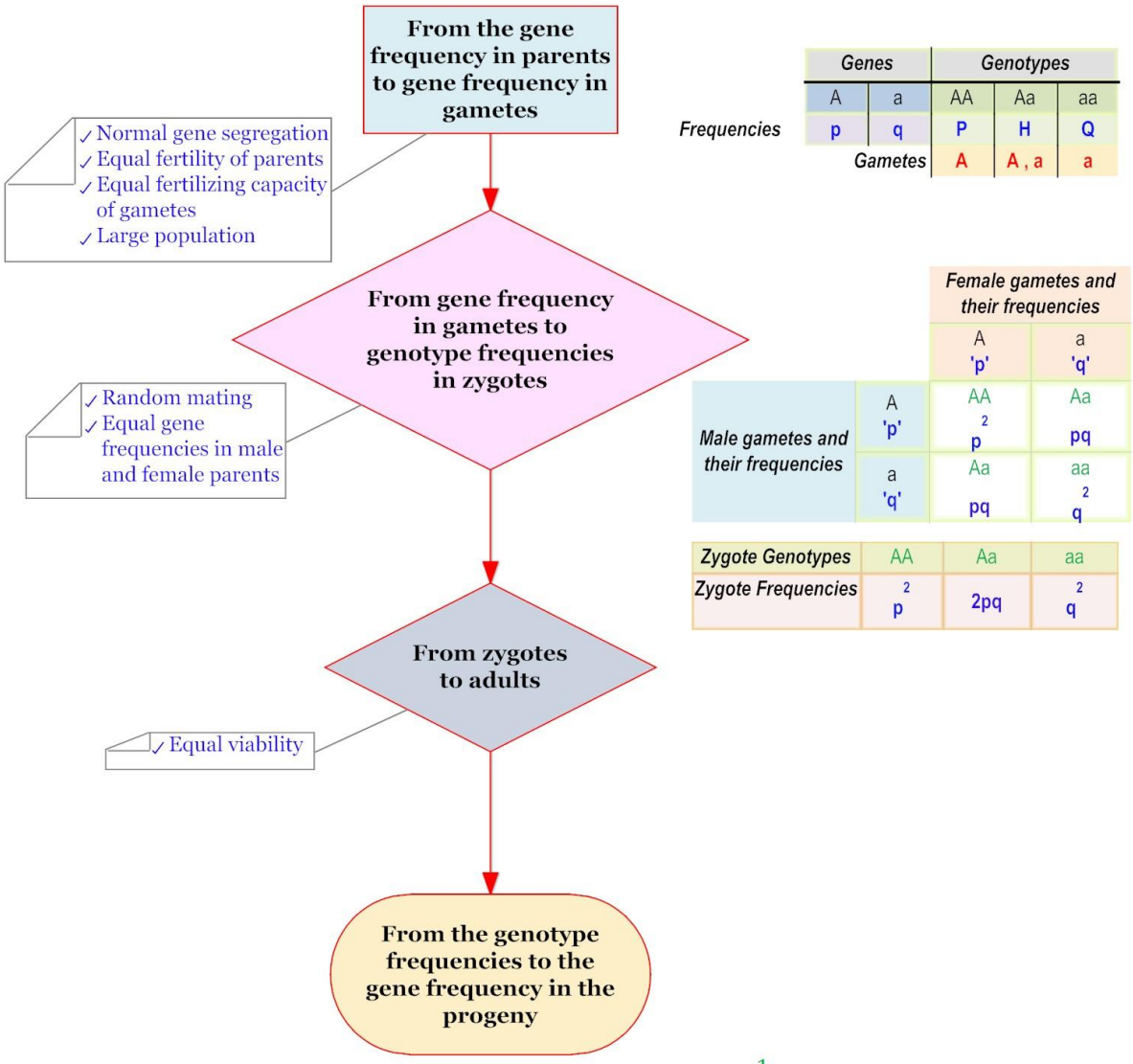
      p = f(A) Frequency of 'A_1' gene
      q = f(a) Frequency of 'A_2' gene

    - Then the next generation will have:
        - The frequency of homozygotes is equal to the gene frequency squared
            - The frequency of the A_1A_1 genotype = p^2
            - The frequency of the A_2A_2 genotype = q^2
        - The frequency of heterozygotes is equal to twice the product of the two gene frequencies
            - The frequency of the A_1A_2 genotype = 2pq
    - For a dimorphic gene the Hardy-Weinberg equation is based on the binomial distribution:
        - $p^2 + 2pq + q^2 = 1$
    - This formula is used to find out the frequency of dominant gene and recessive gene in a population.

p =	Frequency of dominant gene
q =	Frequency of recessive gene
p^2 =	Frequency of dominant homozygote
2pq =	Frequency of heterozygote
q^2 =	Frequency of recessive homozygote

        - Hardy-Weinberg law lays the foundation for the study of population genetics.
- It gives a mathematical approach for genetics and evolution.
- The relationship between gene (allele) frequencies and genotype frequencies expressed by the H-W equation only holds if these 5 conditions are met
    - no new mutations
    - no migration in or out of the population
    - no selection (all genotypes have equal fitness)
    - random mating
    - very large population

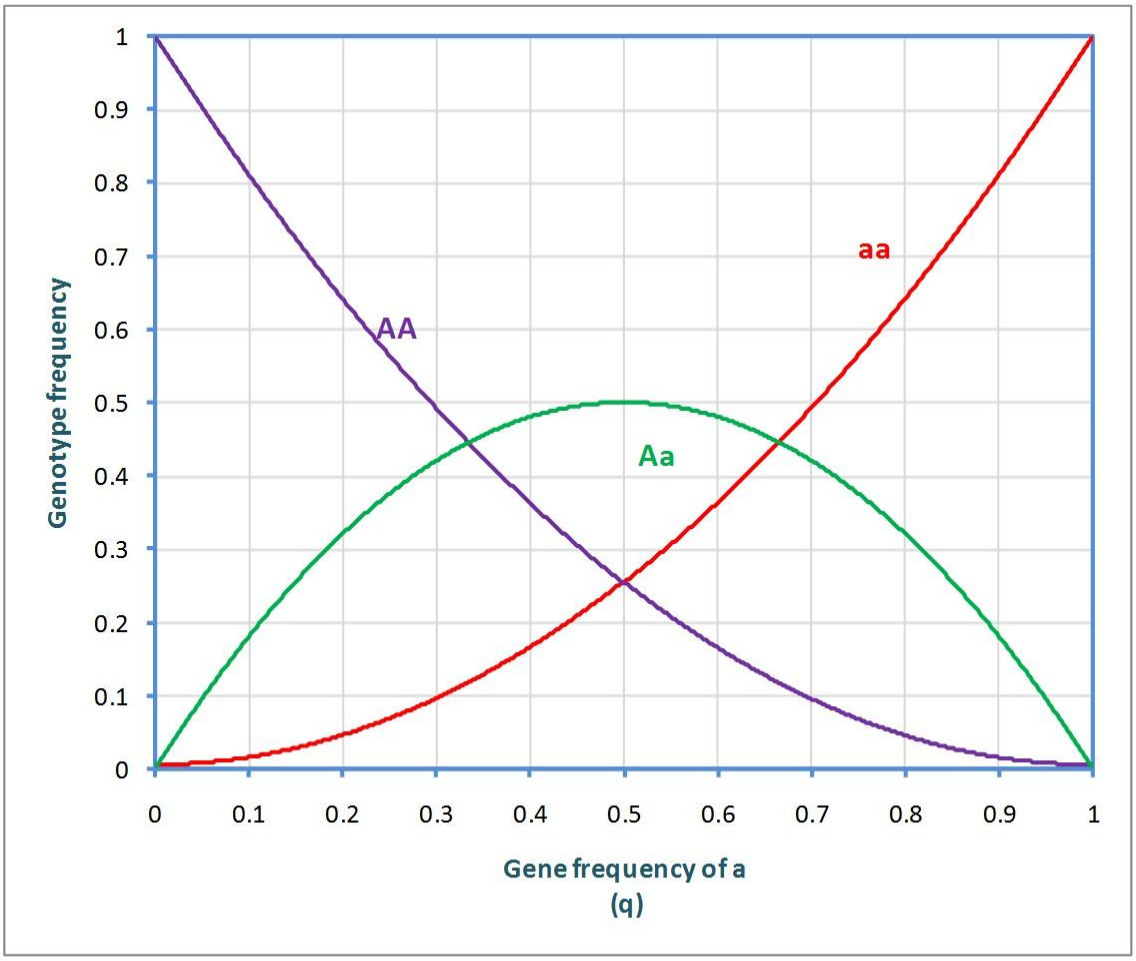
PROOF OF HARDY-WEINBERG LAW



$$Gene\ frequency\ of\ A = p^2 + \frac{1}{2}(2pq)$$
$$= p(p+q) \ \ since\ p+q=1$$
$$= p\ [which\ is\ same\ as\ the\ parent\ generation]$$

$$Gene\ frequency\ of\ a = q^2 + \frac{1}{2}(2pq)$$
$$= q(p+q) \ \ since\ p+q=1$$
$$= q\ [which\ is\ same\ as\ the\ parent\ generation]$$

RELATIONSHIP BETWEEN GENOTYPE AND GENE FREQUENCIES



Relationship between genotype frequencies and gene frequency for two alleles in a population in Hardy-Weinberg equilibrium

- The properties of the population with respect to a single autosomal locus expressed in the Hardy-Weinberg law are:
  - A large random mating population in the absence of migration, mutation and selection is stable with respect to both gene and genotype frequencies.
  - The genotype frequencies in the progeny produced by random mating among the parents are determined solely by the gene frequency among parents.
  - The frequency of homozygotes equals the square of the relevant gene frequency.
  - The frequency of heterozygote equals twice the product of the relevant gene frequencies. Since the frequencies of the genotypes of the homozygotes correspond to the squares of gametic frequencies.
  - Such populations is said to be genetic equilibrium. For a single autosomal locus, the Hardy-Weinberg genotype frequencies are

established by one generation of random mating irrespective of the genotype frequencies among parents.
    - For a single locus with two alleles: the maximum frequency of heterozygote will be 0.5. Then p=q=0.5

APPLICATIONS OF HARDY-WEINBERG LAW

Hardy-Weinberg law has the following applications:

- Calculation of frequencies of recessive and dominant genes in a population
- Calculation of frequency of carriers or heterozygotes in a population
- To test for agreement with a population in Hardy -Weinberg equilibrium

Calculation of frequencies of recessive and dominant genes in a population

- Gene frequencies can be determined from their genotype frequencies, but for this it is necessary to know the frequencies of all three genotypes.
- When there is complete dominance at a locus the expression q = Q + ½ H cannot be used as the classification of genotypes is not possible.
- The heterozygote genotype frequency cannot be estimated, as it cannot be phenotypically distinguished from dominant homozygote.
    - e.g.: Stem length in garden pea plant
        - Tall - dominant (TT, Tt)
        - Dwarf - recessive (tt)
- However, if all the genotypes are in Hardy – Weinberg equilibrium, we need not know the frequencies of all genotypes.
    - Let T - be a dominant gene with a frequency of p and t - be a recessive gene with a frequency of q ; then the frequency of tt homozygote is q^2 and therefore the frequency of recessive allele will be square root of the homozygote frequency.
- For this estimation to be valid there should not be selective elimination of recessive homozygotes.
- Since $p + q = 1$
- p (frequency of dominant allele) = $1 - q$

(TOP)

Calculation of frequency of carriers or heterozygotes in a population

- It is often of interest to know the frequency of heterozygotes or carriers of recessive genes or recessive abnormalities. This can be calculated if the gene frequency is known.
- If Hardy-Weinberg equilibrium is assumed the frequency of heterozygotes among all individuals in the population can be estimated from the formula $2pq = 2q(1-q)$.
    - Example: if q^2 is 0.04 then the gene frequency is q = $(0.04)^{1/2}$ = 0.2

- The frequency of heterozygote is $2q\ (1\text{-}q)$ = 2 × 0.2 × (1-0.2) = 2× 0.2× 0.8 = 0.32
- 0.32 or 32% is of carriers or hererozygotes.

(TOP)

To test for agreement with a population in Hardy -Weinberg equilibrium

- If data are available for a locus where all the genotypes are recognizable then the observed genotype frequencies are used to test for Hardy-Weinberg equilibrium.
- According to the Hardy-Weinberg law, the genotype frequencies of progenies are determined from the gene frequencies of their parents.
- If the population is in H-W equilibrium, frequency is same in progenies as in parents.
- From the gene frequency expected genotype frequencies are calculated. From them the expected numbers are arrived. The agreement between the expected and observed numbers is tested using Chi-square test.
- Example:
  - In a given population of randomly mating gerbils, 500 homozygote brown (BB), 400 heterozygote brown (Bb) and 100 homozygote black (bb) gerbils were observed.
  - Test whether this population is in Hardy-Weinberg equilibrium.
  - NOTE: the black allele (b) is recessive to the wild type brown (agouti) allele (B).

	Genotype				Gene frequency
	BB	Bb	bb	Total	p
bserved	500	400	100	1000	{500+(400x0.5)}/1000 =0.7
xpected	$(0.7)^2$x1000 = 490	(2x0.7x0.3)x1000 = 420	$(0.3)^2$x1000 = 90	1000	
(O –E)	10	-20	10		
	0.204	0.952	1.111		

- Chi-square value = 2.267
- P value = 0.32
- The discrepancy is not significant and could easily have arisen by chance in the sampling.
- We conclude that the genotype frequencies in this population are not significantly different than what would be expected if the population is in Hardy-Weinberg equilibrium

HARDY-WEINBERG LAW-MULTIPLE ALLELES

- If dominance is lacking, calculation of gene frequency is simple in multiple allelic systems.
- The best gene frequency estimate comes from simple gene counting.
- For example, in European cattle breeds, three transferrin alleles A, D and E occur.
- No dominance exists and so six genotypes can be distinguished, i.e., AA, AD, AE, DD, DE and EE.
- If we let these symbols denote the numbers of each genotype and N denotes the total number, we may calculate the gene frequencies as follows:

$$f(A) = p = \frac{2AA + AD + AE}{2N}$$

$$f(D) = q = \frac{2DD + DA + DE}{2N}$$

$$f(E) = r = \frac{2EE + EA + ED}{2N}$$

- For a three allele system, the equilibrium genotype frequencies can be expressed algebraically as p + q + r = 1

$$(p + q + r)^2 = p^2 + q^2 + r^2 + 2pq + 2pr + 2qr = 1$$

- The ABO blood groups in man are determined by a series of three multiple allelic genes: A, B and O alleles.
- If there is dominance, then the estimation is slightly complicated, and approximate methods have to be used.

HARDY-WEINBERG LAW: SEX-LINKED GENES

- In mammals, the female is the homozygous sex, with two X chromosomes (XX), while the male is heterozygous, with one X and one Y chromosome (XY).
- Genes on the X or Y chromosome are called sex linked genes.
- The relationship between gene frequency and genotype frequency in the homogametic sex is the same as with an autosomal gene; but the heterogametic sex has only two genotypes and carries only one gene instead of two.
- So , two-thirds of the sex-linked genes in the population are carried by the homogametic sex and one-third by the heterogametic sex.
- Consider two alleles A and a with frequency p and q

	Female			Male	
Genotype	AA	Aa	aa	A	a
Genotype Frequency	P	H	Q	R	S

- The frequency of A among the females is

$$p_f = P + \frac{1}{2}H$$

- The frequency of A among the male is

$$p_m = R$$

- The frequency of A in the whole populations is

$$\overline{p} = \frac{2}{3}p_f + \frac{1}{3}p_m$$

$$\overline{p} = \frac{1}{3}(2\,p_f + \,p_m)$$

$$\overline{p} = \frac{1}{3}(2P + H + R)$$

- Hence, if gene frequencies among males and among females are different, the population will not be in equilibrium.
- The gene frequency in the population as a whole does not change; but its distribution between the two sexes oscillates as the population approaches equilibrium.
- This oscillation is because of getting sex linked genes in males only from their mother.

$$p_m^{'} = p_f$$

- Females get their sex linked genes equally from both parents

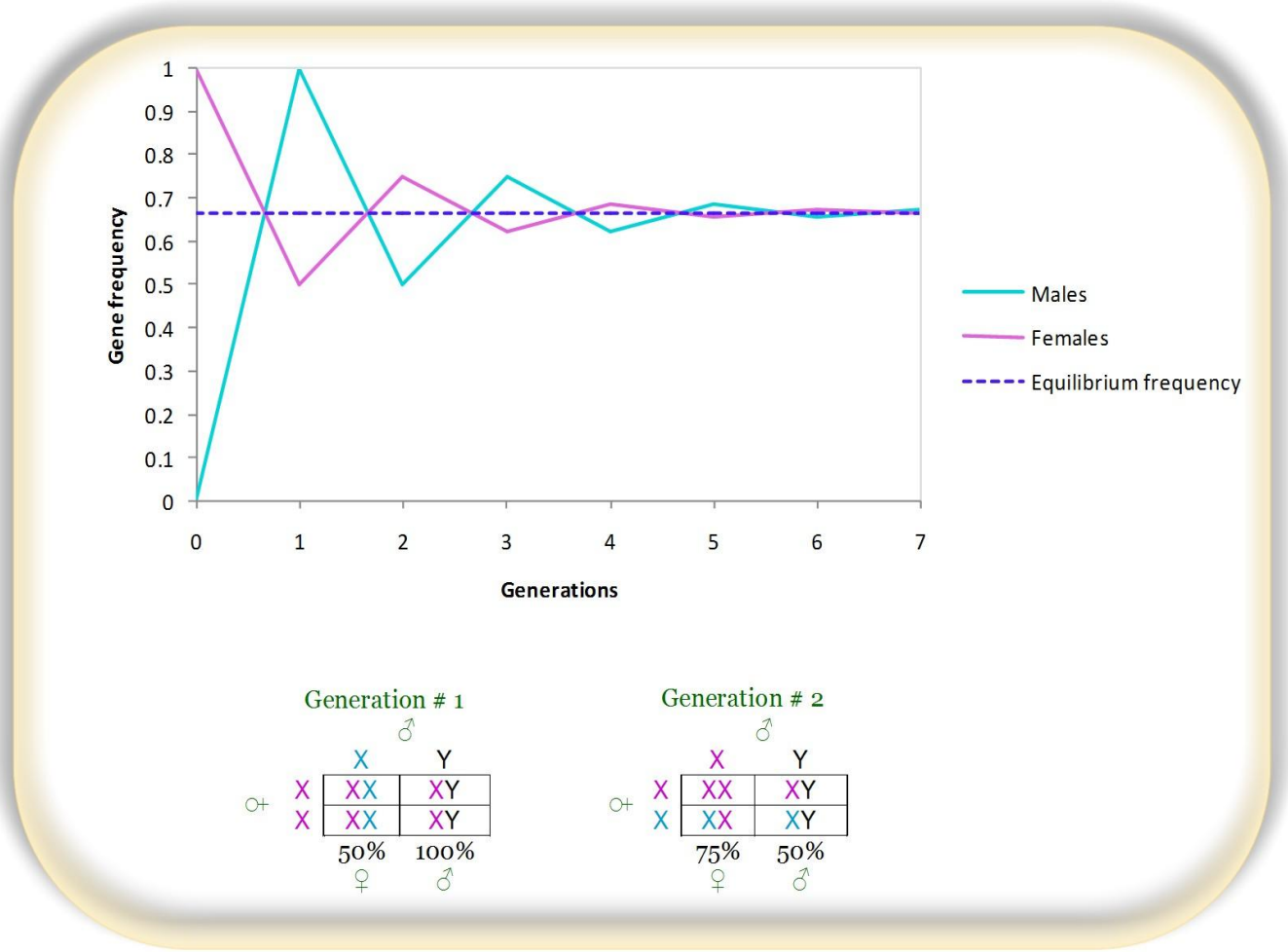
$$p_f^{'} = \frac{1}{2}(\,p_f + \,p_m)$$

- The difference between the frequencies in the two sexes is

$$p_f^{'} - p_m^{'} = \frac{1}{2}(\,p_f + \,p_m) - \,p_f$$

$$p_f' - p_m' = -\frac{1}{2}(p_f - p_m)$$

- It is half the difference in the previous generation. Therefore, if the gene frequencies are different in males and females then one generation of random mating is not sufficient to achieve Hardy-Weinberg equilibrium.
- It may take several generations and the number of generations will depend on the magnitude of difference between the sexes in gene frequency.
- The difference in gene frequency between the sexes will be halved as compared to the previous generation and the sign will be opposite.



- The most important implication is that sex-linked characters are expected to occur with different frequencies in males and females.
- This is relevant to the sex-linked recessive traits for which the frequency of the condition in males (q) is expected to be much higher than the frequency of the conditions in females (q^2).

MORE THAN ONE LOCUS

- The attainment of equilibrium in genotype frequencies after one generation of random mating is true of all autosomal loci considered individually.
- But it is not true of the genotypes considered jointly .
- Consider a population made up of equal numbers of AABB and aabb individuals of both sexes. The gene frequency at both loci is 0.5.

- If the individuals are mated at random the possible genotypes are:

AABB	**AaBB**	**aaBB**
AABb	**AaBb**	**aaBb**
AAbb	**Aabb**	**aabb**

- Only three out of nine genotypes would appear in the progeny in the next generation i.e. the two original homozygotes (AABB and aabb) and the double heterozygote (AaBb).
- The genotype AAbb would be absent though its frequency in equilibrium population would be 1/16.
- The missing genotypes appear in subsequent generations but not immediately at their equilibrium frequency.
- Therefore when two loci are considered together the genotype frequencies will reach equilibrium after several generations of random mating.
- If three loci are considered together, then the number of generations required to reach equilibrium genotype frequencies will be more than that required for two loci considered jointly.

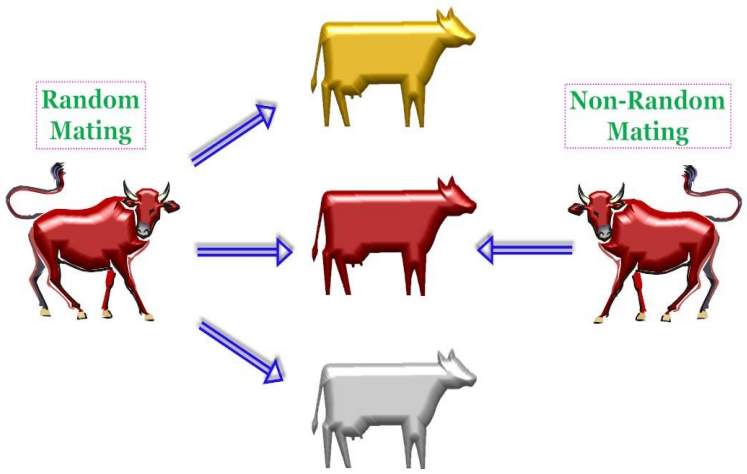
Linked loci

- Under random mating, loci that are linked approach equilibrium more slowly than do loci segregating independently.
- Further more closely the linkage the slower the approach to equilibrium. When equilibrium is reached coupling and repulsion phases are equally frequent:
  - Coupling heterozygotes:
  - AB / ab Repulsion heterozygotes: Ab / aB

FACTORS UPSETING HARDY - WEINBERG EQUILIBRIUM

- This deviation of the equilibrium is brought about by the following evolutionary forces,
  - Mutation.
  - Natural selection.
  - Non-random mating.
  - Genetic drift and
  - Migration and gene flow.

NON-RANDOM MATING



- Nonrandom mating occurs when the probability that two individuals in a population will mate is not the same for all possible pairs of individuals.
- There are two types of non-random mating

Assortative mating

- If mated pairs are of the same phenotype more than would occur by chance this is called assortative mating.
- Mates are genetically similar Example: inbreeding – mating between close relatives includes self-pollination.

Disassortative mating

- Disassortative mating is mating of individuals of different phenotypes.
- Mates are genetically different.

MODULE-23: CHANGE OF GENE AND GENOTYPIC FREQUENCIES

- Learning objective
- After completing this module, the learner should be able to understand the agents that change and affect the genetic makeup of populations.

FORCES OR AGENCIES CHANGING GENE AND GENOTYPE FREQUENCIES

- A large random mating population is stable with respect to gene frequencies and genotype frequencies in the absence of agencies tending to change its genetic properties.

- The shifts or changes in frequencies can be produced by two sorts of process.

Systematic processes

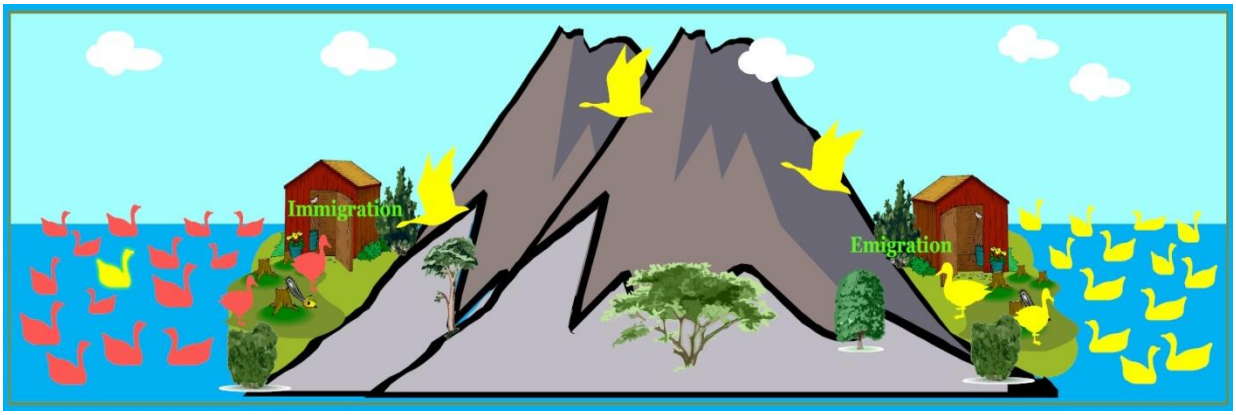
- These tend to change the gene frequency in a manner predictable both in amount and in direction.
- These act both in large and small population.
    - There are three systematic processes :
        - Migration
        - Mutation and
        - Selection.

Dispersive process

- This arises in small populations from the effects of sampling and predictable in amount but not in direction.
- These act only in small population from the effect of sampling.

MIGRATION

- Migration is the movement of individuals from one breeding population to another.
    - *Immigration* is the inward migration of individuals into a population from other populations.
    - *Emigration* is the outward migration of individuals from a population. This brings about the reduction in the size of the gene pool.
- The migration of breeding animals to or from a population can cause changes in gene frequency.



- Let us suppose that a large population consists of a proportion of "m" of new immigrants in one generation then the remainder (1-m) being natives.
  - Number of natives = n_1
  - Number of immigrants = n_2

$$m = \frac{n_2}{n_1 + n_2}$$

$$1 - m = \frac{n_1}{n_1 + n_2}$$

- Let the frequency of a certain allele (A) be q_m among the immigrants and q_o among the natives.
- Then the frequency of the allele in the mixed population q_1 will be

$$q_1 = m\, q_m + (1 - m)\, q_o$$

$$q_1 = m\, q_m + q_o - m\, q_o$$

$$q_1 = m\, q_m - m\, q_o + q_o$$

$$q_1 = m\, (q_m - q_o) + q_o$$

- The gene frequency in mixed population will depend on the original gene frequency of the population and the difference in gene frequency between the immigrants and native ($q_m - q_o$) and the proportion of immigrants.
- The change of gene frequency Δq brought about by one generation of immigration is the difference between the frequency before immigration and the frequency after immigration.

$$\Delta\, q = q_1 - q_o$$

$$\Delta\, q = m\, (q_m - q_o) + q_o - q_o$$

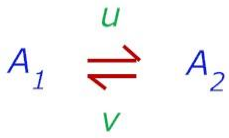
$\Delta q = m (q_m - q_o)$

- Thus the rate of change of gene frequency in a population subject to immigration depends on the immigration rate and the difference in gene frequency between the immigrants and the natives.

MUTATION

- Mutations could lead to occurrence of new alleles and thereby it changes the gene pool of the population.
- It may be favourable or deleterious to the individual's ability to survive.
- If changes are advantageous, then the new alleles will tend to prevail by being selected in the population.

- If a wild allele A_1 mutates to A_2 with a frequency of u per generation.
- u is the proportion of all A_1 alleles that mutate to A_2 between one generation and the next.
- If the frequency of A_1 in one generation is p_o
- Then
  - The frequency of newly mutated gene A_2 in the next generation = $u\ p_o$
  - The new gene frequency of A_1 in the mutated population = $p_o - u\ p_o$
  - Therefore the change of gene frequency = $- u\ p_o$
- Suppose the gene mutates in both directions and the initial allele (gene) frequencies are p (A_1), q (A_2)



- Then the change of gene frequency in one generation
  - $\Delta q = up - vq$
- This situation leads to equilibrium in gene frequency at which no further change takes place. The point of equilibrium can be found by equating the change of frequency Δ q to zero.
  - $pu - qv = o$
  - $qv = pu$
  - $qv = (1-q)u$

- $qv = u - qu$
- $qv + qu = u$
- $q(v+u)=u$
- $q = u / (u + v)$
- Similary $p = v / (v + u)$

- If the mutational rates of A_1 to A_2 (u) and A_2 to A_1 (v) are known at equilibrium then the frequency of A_1 allele and A_2 allele can be calculated directly without using conventional method of estimating gene frequency

SELECTION

- Selection is differential reproduction. It occurs whenever the various kinds of individuals reproduce at different rates.
- Individuals differ in viability and fertility and contribute different number of progeny to the next generation.
- The contribution of offspring to the next generation is called fitness (W) of the individual or adaptive value or selective value.
- Selection favoring certain genotypes should cause alleles to increase in frequency and vice versa.
- The strength of selection is expressed as the *coefficient of selection* "s" which is the proportionate reduction in the gametic contribution of a particular genotype compared with the standard genotype, the usually most favoured one.
- If the fitness of the standard genotype is taken as 1, then the fitness of the genotype selected against is 1 – s.
  - W = 1 - s

Complete selection against dominant gene

- The coefficient of selection is *1* or fitness is *0*
- One generation of selection is sufficient to eliminate all the dominant genes provided there is complete penetrance.
- In the next generation, all the individuals will be of recessive homozygotes and the frequency of recessive allele (q) will be one.

Selection against recessive homozygote (partial selection against recessive)

	Genotypes			
	$A_1 A_1$	$A_1 A_2$	$A_2 A_2$	Total
Initial frequency	p_0^2	$2p_0q_0$	q_0^2	*1*
Coefficient of selection	*0*	*0*	*s*	

Fitness	1	1	1-s	
Gametic contribution	p_o^2	$2p_oq_o$	$q_o^2(1-s)$	$1-s\ q_o^2$

$$q_1 = \frac{q_0 - sq_0^2}{1 - sq_0^2}$$

- The change in gene frequency of a recessive allele as a result of selection

$$\Delta q = q_1 - q_0 = -\frac{sq_0^2(1 - q_0)}{1 - sq_0^2}$$

- Complete selection against recessive: It will not be possible because we can eliminate only those recessive alleles which are present in recessive homozygote, leaving the heterozygote undetected.
- Number of generations required
  - The number of generations required to change the gene frequency from q_o to q_t is

$$t = \frac{1}{q_t} - \frac{1}{q_0}$$

where t = number of generations

q_t is the frequency after t generations of complete elimination of recessives

q_o is the initial (recessive) gene frequency

Selection favouring heterozygote (Overdominance)

- If the fitness of the heterozygote is superior to the respective homozygote then selection will favour heterozygote

	Genotypes			
	A_1A_1	A_1A_2	A_2A_2	Total
Initial frequency	p_o^2	$2p_oq_o$	q_o^2	1
Coefficient of selection	s_1	0	s_2	
Fitness	$1-s_1$	1	$1-s_2$	

Gametic contribution	$p_0^2(1-s_1)$	$2p_0q_0$	$q_0^2(1-s_2)$	$1-s_1p_0^2-s_2q_0^2$

$$\Delta q = \frac{p_0 q_0 (s_1 p_0 - s_2 q_0)}{1 - s_1 p_0^2 - s_2 q_0^2}$$

- When selection favours the heterozygote, the gene frequency of the two alleles A_1 and A_2 tend towards equilibrium at an intermediate value, both alleles remaining in the population.
- The condition for equilibrium is that $\Delta q = 0$ and is fulfilled in generation when $s_1 p = s_2 q$

$$q = \frac{s_1}{s_1 + s_2} \text{ and similary } p = \frac{s_2}{s_1 + s_2}$$

DISPERSIVE PROCESS

- Dispersive process differs from systematic processes in being random in direction and predictable only in amount.
- If systematic factors were present, as in very large population, gene frequencies would reach equilibrium and remain there until external conditions change.
- This property of stability does not hold in small populations and the gene frequencies are subject to random fluctuations arising from sampling of gametes.
- The gametes that transmit genes to the next generation carry a sample of the genes in the parent generation.
- If the sample is not large, the gene frequencies are liable to change from one generation to the next.

Causes

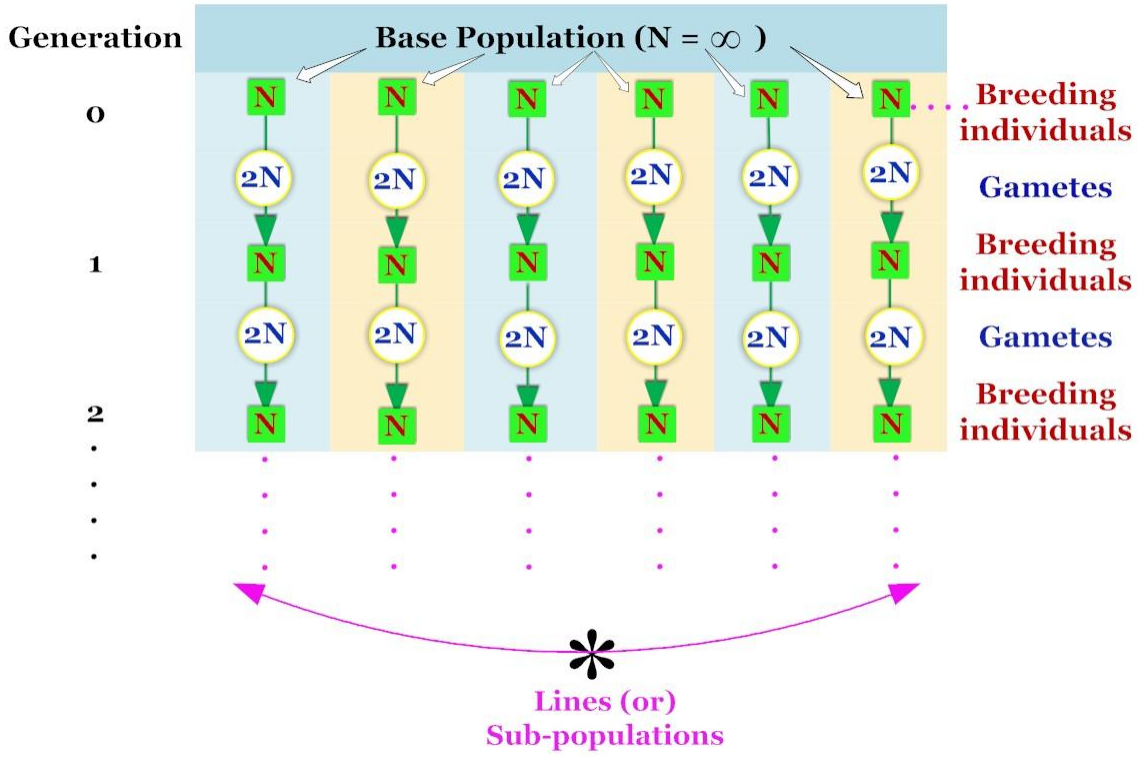
- Small population size
- Founder effects – occurs when a population is initially established by small number of breeding individuals
- Bottleneck effect – occurs when a population is dramatically reduced in size

Effects

- Random drift
- Differentiation between sub-populations
- Uniformity within sub-populations
- Increased homozygosity

THE IDEALIZED POPULATION

- The Idealized Population In order to deduce the dispersive process to its simplest form we imagine an idealized population as follows:
  - Suppose there is one large population in which mating is random, and this population becomes sub divided into a number of sub-populations.
  - The initial random mating populations will be referred as the base population and the sub-populations will be referred to as lines.
  - Each line is considered as a small population in which gene frequencies are subject to the dispersive process.
- Conditions for the idealized population are as follows:
  - The generations are distinct and do not overlap.
  - Mating is restricted to members of the same line or in other words migration is excluded.
  - The number of breeding individuals is equal for all lines and in all generations.
  - Mating is random within each line.
  - Selection is absent.
  - Mutation is disregarded.



- The conditions specified for the idealized population may not hold in real population.

VARIANCE OF GENE FREQUENCY

- The change of gene frequency Δq resulting from sampling in one generation can be stated in terms of its variance
    - This variance of Δq expresses the magnitude of the change of gene frequency resulting from dispersive process.
    - In the next generation, the sampling process is repeated starting from the gene frequency in the previous generation.
    - The effect of continued sampling through successive generations is that each line fluctuates irregularly in gene frequency and the lines spread apart progressively, thus becoming differentiated.
    - Sooner or later each line must reach the limits of *0* or *1*.
    - When a particular allele frequency has reached *1* it is said to be fixed in that line and if it reaches *0* it is lost.
    - When a particular allele is fixed in all individuals in a line; all are identical for that genotype and become genetically identical.
    - This is the basis of the genetic uniformity of highly inbred strains.

$$\sigma^2_{\Delta q} = \frac{pq}{2N}$$

where N - subpopulation or line size (sample)

INBREEDING

Inbreeding means the mating together of individuals that are related to each other by ancestry

- Pairs mating at random are more closely related to each other in a small population than in a large one.
- Hence, the properties of small population can be treated as the consequence of inbreeding.
- So the degree of relationship between the individuals in a population depends on the size of the population.
- Genes may be similar or identical due to two different reasons
- Homozygotes – *Identical by descent*
    - Two genes are identical by descent (autozygous) if they are biochemical replicates produced without mutation from a common ancestral gene.
- Homozygotes - *Alike in state (independent/ allozygous)*
    - Two genes are regarded as alike because of their nucleotide sequence and similar function.

Inbreeding coefficient

- Inbreeding coefficient is probability that the two genes at any locus in an individual are identical by descent.
- The symbol for coefficient inbreeding is *F*.

Panmictic index is the probability that genes at any single locus are independent by descent P = (1 – F)

- The inbreeding coefficient of subsequent generations expresses the amount of dispersive process that has taken place since the formation of the base population.
- The amount of dispersive process expressed in terms of increment of inbreeding or rate of inbreeding is $\Delta F = 1/2N$
  - Where ΔF is the increment in inbreeding in each generation and N is the number of breeding individuals in the population.
- Therefore, the increment of inbreeding can be used instead of the population size to estimate the variance of change of gene frequency

$$\sigma^2_{\Delta q} = \frac{pq}{2N} = pq\Delta F$$

EFFECTIVE POPULATION SIZE

- In domestic animals, the sexes are often unequally represented among breeding individuals and, in general, fewer males than females are used.

enetically Effective Population Size: "The number of breeding individuals in an idealized population that would sho same amount of dispersion of allele frequencies under random genetic drift or the same amount of inbreeding as population under consideration"

- The effective population size is based on the number of genes in the population that can be passed on to the next generation. The symbol is N_e (N-effective)
- N_e the harmonic mean of the two sexes

$$\frac{4}{N_e} = \frac{1}{N_{males}} + \frac{1}{N_{females}}$$

$$N_e = \frac{4N_m N_f}{N_m + N_f}$$

Then the rate of inbreeding is

$$\Delta F = \frac{1}{8N_m} + \frac{1}{8N_f}$$

- Rate of inbreeding is inversely proportional to the effective population size.
- That is, the smaller the effective population size, the greater the increase in inbreeding per generation.
- Thus increase in the number of homozygote in the smaller population.