

Population Genetics

Trait :- Any measurable or observable property of an individual which exhibit inheritable variation in population.

2 types - ✓

Qualitative Trait

(i) Controlling Gene

One or few
(Major genes)
or Oligogenes

(ii) Environmental Effect

Little / No effect

(iii) Distribution

Binomial or
Poisson's dist.

(iv) Variations

Discontinuous Variations

(v) Data Recording

By observation / Counting

(vi) Analysis

Ratio, Proportion
Percentage

Quantitative Trait

Many genes
(minor genes / Polygenes).
↓
given by math

Much affected by environment

Normal distribution

Continuous Variations

By measurement

Mean, Variance,
Co-variance

Study of genetic variation within or between the population that focusing on how allelic and genotypic frequencies changes over time due to evolutionary processes → Population genetics.

→ Population :- Study of the qualitative trait of the Genetics level of population and changes over the generation is k/a population genetics.

Genotype :- It is the proportion or % of a particular genotype among all the individuals in the population.

e.g. There are 25 individuals or genotypes as AA in a population of 100 individuals
then G.F. = $\frac{25}{100} = 0.25$

$$\text{G.F.} = \frac{\text{No. of individuals of a particular genotype}}{\text{No. of total individuals in population}}$$

Allele/Gene :- It is the proportion or % of a particular allele among all the alleles present at their locus in the population.

→ The Sum of all allele frequencies will be 1.

$$\text{e.g. } A_1A_1 = 30$$

$$A_1A_2 = 50$$

$$A_2A_2 = 20$$

100

For a normal gene having 2 allele in population individual The G.F. are denoted by P, H, Q

$$A_1A_1 \rightarrow 30 \times 2 = 60$$

$$A_1A_2 \rightarrow 50$$

$$A_2A_2 \rightarrow 20 \quad \frac{110}{200}$$

$$\begin{aligned} & \left. \begin{array}{l} \text{dominant homozygote} \\ \text{Recessive homozygote} \end{array} \right\} \begin{array}{l} A_1 = 60 \\ A_2 = 50 \end{array} \\ & \left. \begin{array}{l} \text{dominant homozygote} \\ \text{Recessive homozygote} \end{array} \right\} \begin{array}{l} A_1 = 50 \\ A_2 = 40 \end{array} \\ & \left. \begin{array}{l} \text{dominant homozygote} \\ \text{Recessive homozygote} \end{array} \right\} \begin{array}{l} A_1 = 90 \\ A_2 = 90/200 \end{array} \end{aligned}$$

The Genotype freq. are denoted by P, H, Q for dominant, Heterozygote and recessive homozygote.

p → dominant allele

q → recessive allele

$$[P + H + Q = 1]$$

$$[p + q = 1]$$

Hardy-Weinberg Law ✓

Given by G.H. Hardy (A British mathematician) & W. Weinberg (A German Physician) in 1908 independently.

In a large random mating population, the gene & genotype frequencies are constant from generation to generation in the absence of mutation, migration & selection.

The population with constant gene & genotype freq. is said to be in Hardy-Weinberg equilibrium.

There is a simple relationship b/w gene & G.F. that if the G.F. of 2 alleles among the parents are p & q, then the G.F. among the progeny are $p^2, 2pq, q^2$

* Conditions for H.W. law that must be followed.

1. Large population :- The number of adult individuals must be in hundreds rather than in tens.

2. Random mating :- thousands

Mating within the population for the genotypes under consideration occur at random.
(Panmictic population)

or germplasm

3. No mutation (no new allele is created, no allele is lost)
4. No migration (There is no movement of individuals from or into the population (i.e. a closed population) in the form of live animals or frozen embryos or gamete.)
5. G.F. must be same in ♂ & ♀
6. There should be no linkage.

7. Equal fertility of the parents & Equal survivability of all zygotes means there should be no selection either naturally or artificially.

* Gene pool :- The sum total of all the genes possessed by a population.

Genotype	No. of animals	Genotypic fr.	Number of alleles
AA	35	$35/100 = 0.35$	A a
Aa	50	$50/100 = 0.50$	$35 \times 2 = 70$
aa	15	$15/100 = 0.15$	$50 \times 1 = 50$
Total	100	<u>1.00</u>	$-$
			$15 \times 2 = 30$
			120
			$\frac{80}{80}$

$$f(A) = \frac{120}{200} = 0.6$$

$$f(a) = \frac{80}{200} = 0.4$$

$$f(A) + f(a) = 0.6 + 0.4 = 1$$

$$\left\{ \begin{array}{l} p = P + \frac{1}{2} H \\ q = Q + \frac{1}{2} H \end{array} \right.$$

★★ If a population is in HW-equilibrium then

$$\left\{ \begin{array}{l} P = p^2 \\ Q = q^2 \\ H = 2pq \end{array} \right.$$

Mendelian Genetics

- Deals with study of pattern of transmission of genes from parents to progeny in a single family/cross pedigree.
- Unit of study or organization is a family or an individual.
- Specific matings are followed from parents to progeny generation in terms of genotypic and phenotypic ratio.

Population Genetics

- Deals with study of genes transmission from parent to progeny by all the members (i.e. all families) of a mendelian population.
- Unit of study or organization is population.

→ Evolution involves progressive changes in the genetic makeup of populations brought about by evolutionary agents acting over a long period of time.

→ Breeders of domestic species also make efforts to improve the performance of animals by alteration of genetic makeup of their herds through selection, introduction of new genes.
Factors → that changes the allelic fr.

1. Systematic forces (directional changes) :-

→ Predictable both in amount and direction.

→ Migration, mutation and ^{selection} fall under this category b/c they operate in a directional manner.

2. Dispersive (random changes) :- The force which changes the allelic frequency in a manner that -

Predictable in amount but not in direction.

* We shall examine the effects of each of these forces separately assuming that only one force is operating at a time.

(A)

Mutation

e.g. Random genetic drift

Sudden & Permanent alteration in a gene giving rise to a new allelic state of that gene.
(Unidirectional)

• Non-recurrent v/s Recurrent mutation

Recurrent mutation is the one that occurs at a certain regular rate in every generation.

& is of ^{have} importance in changing the gene frequency.
e.g. polledness allele that occurred in several cattle.

Random → Panmixia
Mating

Non-recurrent mutation :- A mutation that occurs as a rare and unique event and thus gives rise to just one representative form of the mutated gene.

Mutant alleles resulting from non-recurrent mutations rarely become established in a population and majority of these are lost within a few generations of their occurrence due to chance or selection.

It is of little importance as a cause of change in gene frequency.

Forward v/s Backward mutation

Forward mutation changes the allelic state from the original (wild type) to an abnormal (mutant) form.

Since most of the wild forms are dominant, forward mutations usually change a gene from dominant to recessive state.

Reverse or back mutation is the one that changes the mutant form of allele to standard wild type form (usually from recessive to dominant form).

Reverse mutation thus restores the original form of allele.

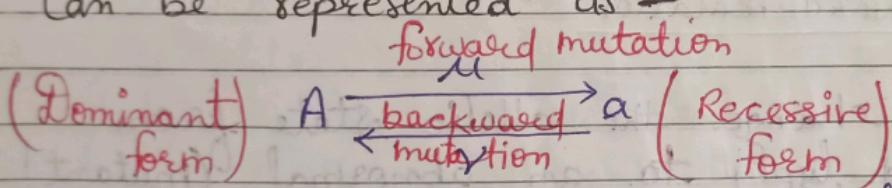
Mutation Rate :-

→ Mutation rate is the proportion with which a particular allele changes to an alternative form.

• Mutation rate is expressed on per gamete or per-cell cycle/generation - basis.

• ~~Mutation Rate~~ = $\frac{\text{Number of mutant alleles}}{\text{Total number of alleles of that gene in the population.}}$

The process of forward and backward mutations can be represented as -



Forward mutation will reduce the fr. of 'A' and increase the fr. of 'a'

Change in fr. of 'a' (Δq) following one generation of forward mutation will be

$$\Delta q = +p$$

$p \rightarrow$ initial fr. of allele A

$q \rightarrow$ initial fr. of allele a

$\mu \rightarrow$ forward mutation rate (from A to a)

$\gamma \rightarrow$ reverse mutation rate (from a to A)

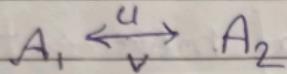
Backward mutation from a to A will tend to increase the fr. of A and decrease the fr. of a.

Change in fr. of a (Δq) due to backward mutation will be $\Delta q = -\gamma p$.

The net change in fr. of a allele due to forward and backward mutation will be -

$$\Delta q = u\bar{p} - v\bar{q}$$

→ The rate of change in allelic fr. due to mutation thus depends upon initial allelic frequencies and mutation rates (forward & backward).



$$\frac{p}{q} \quad q_0$$

→ Next gen. (after mutation)

$$\begin{aligned} \bar{p}_1 &= \bar{p}_0 + v\bar{q}_0 - u\bar{p}_0 \\ \Delta q &= \bar{q}_1 - \bar{q}_0 \\ \Delta q &= u\bar{p}_0 - v\bar{q}_0 \end{aligned}$$

At eq^m Forward mutation = Backward mutation

$$\Delta q = 0$$

$$u\bar{p}_0 = v\bar{q}_0$$

$$u\bar{p}_0 = v\bar{q}$$

$$u(1-q) = vq$$

$$u - uq = vq$$

$$u = uq + vq$$

$$u = q(u+v)$$

$$q = u/u + v$$

→ Rate of mutation is very low 10^{-5} or 10^{-6} per generation.

→ Reverse mutation is less frequent than forward mutation.

(B) Migration

→ It refers to movement of individuals from one breeding population into another where they interbreed and become a part of gene pool.

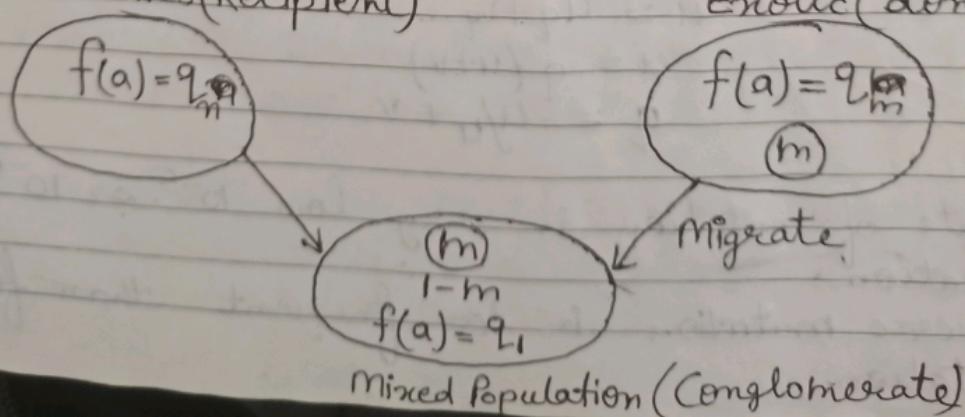
- Individuals moving out of a population → Emigrants
- Individuals moving into a population → Immigrants

* Migration is not simply the physical movement of animals but the migrating animals must participate in matings and contribute their genes to gene pool of future generations of recipient population.

* A single introduction of individuals into a new population is k/a gene exchange

* If the migration is constant and recurrent it is known as gene flow.

To examine the effect of migration on change in allelic frequency, consider two populations designated as donor (exotic) and recipient (native).
Natives (Recipient) Exotic (donor)



q_m = fr. of recessive allele in migrants/ donors
 q_n = fr. of recessive allele in natives/ recipients
 q_i = fr. of recessive allele in mixed population (Conglomerate)

N_n = number of native (recipient) individual

N_m = number of migrants (donor) individual

m = proportion of migrants in the mixed population and is commonly k/a migration or immigration coefficient.

Migration Coefficient is a measure of the proportion of alleles contributed by migrants into the new mixed population and is calculated as-

$m = \frac{\text{number of donors in mixed population}}{\text{total number in mixed population}}$

$$m = \frac{N_m}{N_n + N_m}$$

$1-m$ = proportion of natives in mixed population

$$1-m = \frac{N_n}{N_n + N_m}$$

Sum of natives and immigrants $[m + (1-m)]$ thus equals one.

Extent of migration

The extent of migration can vary from a single introduction of exotics (donor) into a native population to the complete replacement of natives with the genes of exotic population.

Single introduction of migrants

When a single introduction of exotic animals into a local population, the new mixed population will not be in HW equilibrium. However, a single generation of random mating will establish HW equilibrium. G.F. in the new population will remain unchanged provided other forces are not operative.

Complete replacement of native population

This would require a number of repeated introductions of exotics into the native population (Gene flow.)

The native population will become like exotic.
This is an extreme case & practically not feasible.

examples of migration in livestock

- (i) Introduction of Zebu Cattle in USA
- (ii) Introduction of Danish Landrace hogs in USA
- (iii) Introduction of exotic Cattle in India
- (iv) Replacement of native chickens by exotic Stocks.

② Selection

When individuals of certain genotypes produce more offspring than those of others, selection is said to be operating. In other words, Selection is present when the number of progeny produced by different genotypes is not proportional to their frequencies in population.

2 types

- (i) Natural Selection (ii) Artificial Selection

(i) Natural Selection

It is defined as non-random differential survival and reproduction of different genotypes.

Individuals in a population differ from one another with respect to survival in a given environment.

→ A/c to Darwin and Wallace's theory (1859) natural selection has been the most important force in evolutionary changes in population.

→ Natural selection is the net result of cumulative action of a number of forces (fluctuations in temp., humidity and day length, availability of food.)

→ Natural Selection (given by Darwin) rests on 3 premises.

- (i) Competition for Survival
- (ii) Presence of Variability
- (iii) Survival of fittest

→ Net result of natural selection is to make the population better adapted to its prevailing environment.

Fitness and its Components

Natural selection is mediated through differential viability and/or differential reproductive ability of different genotypes and can operate at any stage of life cycle of an organism.

* The capacity of a given genotype to survive and reproduce under a specific environment, is self referred to its fitness.

The word fitness was used by Darwin to indicate the survival and reproductive ability of a genotype (referred to as Darwin fitness) or reproductive fitness.

Fitness has 2 Components

① Viability - Ability of a newly formed zygote of a specified genotype to survive to the reproductive stage.

② Reproductive ability - Ability of an individual of a given genotype to produce offspring during the reproductive period.

These 2 components determine how many offspring a particular genotype would contribute to next generation.

Examples of Natural Selection

- (i) Industrial Melanism
- (ii) DDT resistance in flies and mosquitoes
- (iii) Heat and disease tolerance in Zebu cattle.
- (iv) Trichostrongyle worm resistance in sheep.

→ Natural selection is very slow process but progressive over a period of time it can bring about large changes in the genetic composition of populations.

- Fitness (w) is the proportionate contribution of progeny to the next generation by a particular genotype as compared to fit genotype.

✓

- Selection coefficient (s) is an indicator of strength of selection and the proportionate reduction in genetic contribution of a particular genotype as compared to most fit genotype]

- fitness is referred to as relative fitness.
- Selection coefficient is a measure of the degree to which a genotype is selected against, it is opposite of fitness.

$$\therefore W = 1 - s$$
$$s = 1 - W$$

• W & s vary b/w zero and one

eg. if $s = 0.1$ then $w = 0.9$ $(W) = 1 - s$

- \rightarrow A genotype that is lethal/sterile and hence fails to contribute any offspring has $s = 1$ and $w = 0$

- \rightarrow A genotype that is most viable and fertile and hence contributes maximum number of offspring has $s = 0$ and $w = 1$

- * Mutation by itself cannot bring about large changes in gene frequency but it is the ultimate source of a gene creation of new alleles in populations. Mutation by creating alternative forms of a gene provides opportunity for selection

- \rightarrow If the value of ' s ' for a genotype is 0.2, it means 20% individuals of that genotype will not be allowed to breed.

Artificial Selection

- Artificial selection is much more rapid than natural selection.
- The definite differences in shape, size, color, growth, production, behaviour pattern among breeds of livestock and companion animals resulted from continued ~~are~~ artificial selection.

Natural Selection

- (i) Caused by force of nature
- (ii) Survival of fittest is main force
- (iii) Very slow process and usually takes 100/1000 of generations to produce meaningful effects.
- (iv) Can occur at any stage of life (from zygote to adulthood)
- (v) Ex. Origin & development of new species of plants & animals

Artificial Selection

- (i) by efforts of man
- (ii) Standards laid down by man, which are usually economic in nature are determining factors.
- (iii) Relatively fast
- (iv) Can occur only when the trait of interest has been recorded & sexual maturity attained
- (v) Ex. Development of new varieties/breeds of animals and plants suited for economic uses.

Genetic effects of Selection

Selection by itself doesn't create new allele gene but by eliminating undesirable genotypes from the population, it causes a change in the frequency of different allele.

$$\text{eg. } f(AA) = 0.25$$

$$f(Aa) = 0.50$$

$$f(aa) = 0.25$$

$$f(A) = 0.5$$

$$f(a) = 0.5$$

If individuals of aa genotype are unable or not allowed to produce progeny. (i.e. that selection is directed against 'a')

Only two genotypes AA and Aa will produce progeny

$$f(A) = \frac{f(AA) + \frac{1}{2}f(Aa)}{f(AA) + f(Aa) + f(aa)} = \frac{0.25 + \frac{1}{2} \times 0.50}{0.25 + 0.50} = 0.67$$

$$\therefore f(A) \text{ in selected group} = 0.67$$

$$f(a) \text{ in " } = 0.33$$

(♀)	A (0.67)	a (0.33)	(♂)
A (0.67)	AA (0.45)	Aa (0.22)	
a (0.33)	Aa (0.22)	aa (0.11)	

$$f(AA) = 0.45$$
$$f(Aa) = 0.22 + 0.22$$
$$= 0.44$$

$$f(aa) = 0.11$$
$$f(A) = 0.67$$
$$f(a) = 0.33$$

$$f(a)_{\text{fr}} = 0.5 \rightarrow f(a)_{\text{fr}} = 0.33$$
$$f(A)_{\text{fr}} = 0.5 \rightarrow f(A)_{\text{fr}} = 0.67$$

Due to selection against homozygous recessive genotype fr. of recessive allele decreased and fr. of dominant allele increased. As a consequence of change in allelic fr., genotypic fr. in population also change.

When selection against aa genotype is continued generation after generation the fr. of a would go on ^{fixation} decreasing.

It increases the fr. of A allele to the point where it becomes the only allele at this locus in the population.

When a particular allele has reached a fr. of one, it is said to have been fixed or reached fixation in that population.

While its fr. becomes zero, it is said to have been lost.

Gene fixation is the point at which a particular allele becomes the only allele at that locus in the population. When an allele is fixed all the individuals in that population would be homozygous for that locus.

w = fitness value

Effectiveness of Selection

A. Partial Selection against a Recessive Gene under complete dominance :-

→ Only a fraction of recessive individuals is rejected

(f_i); for allele $A = p$ [before selection]

(f_i); for allele $a = q$ [in HW-equilibrium]

$s \rightarrow$ Selection coefficient against aa

∴ Dominance is complete ∵ AA, Aa are equally fit

$$(w = 1 \& s = 0)$$

Genotype	f_i	s	Relative fitness (w)	Gametic Contribution (fr. x fitness)
AA	p^2	0	$1-0=1$	p^2
Aa	$2pq$	0	$1-0=1$	$2pq$
aa	q^2	s	$1-s$	$q^2(1-s)$
Sum	1	-	-	$1-sq^2$

∴ Selection against aa will reduce its proportionate gametic contribution and only a fraction of this genotype will contribute to the gene pool of next generation.

Since no selection is practiced against AA and Aa their gametic contribution will be in proportion to their frs. in population whereas the contribution of aa genotype become reduced. $(1-s)q^2$

$$W \text{ (mean fitness)} = p^2 + 2pq + (1-s)q^2$$

$$\boxed{W = 1 - sq^2}$$

$$q \rightarrow (f_i)_a \quad q_i \rightarrow (f)_a \text{ after selection}$$

before selection

$$(i) f(AA) = \frac{h^2}{1-Sq^2}, \quad f(Aa) = \frac{2hq}{1-Sq^2}, \quad f(aa) = \frac{q^2(1-s)}{1-Sq^2}$$

(ii) $\therefore f(AA) + f(Aa) + f(aa) = 1$ (before selection)

$$f(a) \text{ or } q_s = \frac{\frac{1}{2}f(Aa) + f(aa)}{f(AA) + f(Aa) + f(aa)} = \frac{\frac{1}{2} \left(\frac{2hq}{1-sq^2} \right) + \frac{q^2(1-s)}{1-sq^2}}{\frac{p^2}{1-sq^2} + \frac{2hq}{1-sq^2} + \frac{q^2(1-s)}{1-sq^2}}$$

$$F_{q_1} = \frac{1}{2} \times 2fq + q^2(1-s)$$

$$p^2 + 2pq + q^2(1-s)$$

$$q_1 = \frac{1/2 \times 2pq + q^2(1-s)}{p^2 + 2pq + q^2(1-s)} = \frac{1}{1 - Sq^2}$$

24 1 - Sq2

$$q_1 = \frac{hq + q^2(1-s)}{1 - sq^2}$$

$$\therefore p+q = 1$$

$$p = 1 - q$$

1

-2

change in the fr. of recessive allele Aq

$$\Delta q = q_2 - q_1$$

$$\Delta q_1 = \frac{q - Sq^2}{1 - Sq^2} - q$$

$$4q = - \left[\frac{sq^2}{1-sq^2} \right]$$

$$\frac{hq + q^2 - Sq^2}{1 - Sq}$$

$$-\textcircled{3} \quad q \left(k + \frac{q}{s} \right) - s q^2$$

$$\frac{q - \sqrt{q^2}}{1 - \sqrt{q^2}}$$

- Selection is most eff. at intermediate gene fr. and least eff. when q is either large or small
- Selection for or against recessive allele gene is extremely ineffective when recessive allele is rare.
- The effectiveness of selection in changing the allelic fr. depends upon f_i (p & q) and s
- \ominus sign indicates that selection against recessive genotype would decrease the fr. of recessive allele.

$\therefore -Sq^2$ in denominator represents the proportion of homozygote recessives which is lost through selection while $1-Sq^2$ represents the proportion of those who contribute genes to next generation.

$$\therefore \boxed{1q = -spq^2} \quad (1q \propto s)$$

B. Complete Selection against recessive : Number of generations required

- Homozygote recessives (aa) don't produce any progeny.
- e.g. Cases in which recessive genotype causes lethality or sterility

$$\therefore \left\{ \begin{array}{l} q_1 = q - Sq^2 \\ 1 - Sq^2 \end{array} \right\} \text{ from equation } ②$$

\therefore for complete selection, against homozygote recessive allele $S=1$

$$\therefore q_1 = \frac{q - q^2}{1 - q^2} \Rightarrow \frac{q(1-q)}{(1+q)(1-q)} = \frac{q}{(1+q)}$$

$$\boxed{q_1 = \frac{q}{(1+q)}} - ④$$

if we take $q = q_0$ (to indicate generation number)

$$q_1 = \frac{q_0}{1+q_0}$$

$$q_2 = \frac{q_1}{1+q_1} \text{ or } \frac{q_0}{1+2q_0}$$

fr. of recessive allele
in first generation

(f) a in 2nd generation

* Frequency of recessive allele after t generations (q_t)
of complete selection -

$$q_t = \frac{q_0}{1+tq_0} \quad \checkmark$$

* The number of generations (t) required to bring about a specific change in fr. of a recessive allele from its initial value (q_0) to any desired value (q_t)

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

* Number of generations required halving the gene fr. ($t_{1/2}$) from its current value q_0 -

$$t_{1/2} = \frac{1}{q_{1/2}} - \frac{1}{q_0} = \frac{1}{q_0} \quad \checkmark$$

→ It is very difficult to remove a harmful recessive allele from a population b/c it occurs at very low fr. and protected from selection by the heterozygotes.

* [decrease in fr. of α s]
[recessive allele]

Effectiveness of selection against the recessive allele decreases as its fr. becomes very low.

At low fr. a large proportion of recessive alleles are hidden from the action of selection as they occur in heterozygotes.

c. Complete Selection against dominant gene

The change in fr. of dominant gene caused by selection against it -

$$\Delta p = - \left[\frac{spq^2}{1-s+sq^2} \right] \quad \checkmark$$

if selection against A is complete (i.e. $s=1$)

$$\Delta p = -p$$

$p = (\text{f})$; of dominant allele.
 Δp = change in fr. of dominant gene after one generation

The fr. of a dominant allele responds more quickly to selection than recessive allele b/c dominant alleles are exposed to

selection whereas recessive are protected from selection as they are carried by heterozygotes.

e.g. Huntington's chorea is a hereditary disease governed by a dominant autosomal gene.

* There are 2 aspects of hardy - weinberg law

1. The relationship b/w gene frs and genotype frs applies to a single generation.

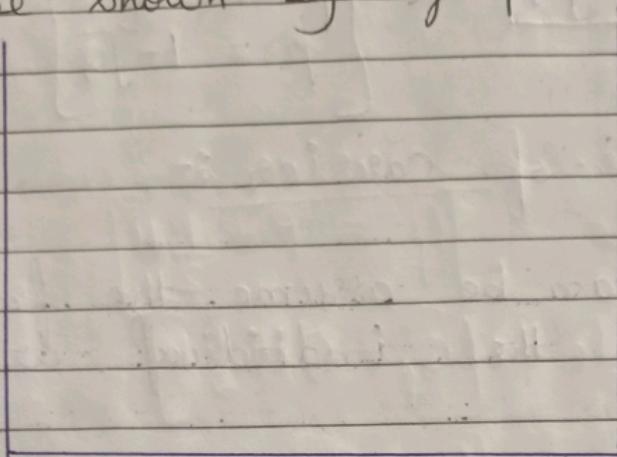
2. The genotype frs in progeny depend only on the gene frs in parents and not on the genotype frs.

→ Parents with any genotype frs provided the mating at random and provided the gene frs in ♂ & ♀ are same, produce progeny in HW - proportion.

→ If the gene fr. is not equal in males and females and than in the 1st generations the frs will be equal in both sexes and the 2nd generation v produce of random mating HW - genotype frequencies.

If the gene frs are same in both ♂ & ♀

Relationship b/w gene fr. and genotype fr. in population in H - w - equilibrium can be shown by graph.



2 Conclusions -

- i) The fr. of heterozygote can't be greater than 50% or 0.5 and the max. occur when $p = q = 0.5$
 $\therefore \{ H = 2pq \}$
- ii) When the gene fr. of an allele is low, the rare allele occurs predominantly in heterozygote and there are very few homozygotes.

Applications of H - w - law :-

- i) To know the gene fr. of recessive allele.
In case of complete dominance the heterozygote is indistinguishable. The heterozygote have the phenotype as the dominant genotype.
- If the genotypes are in H - w - proportion then we first calculate the gene fr. of recessive allele, that is square root of

genotype fr. of recessive genotype.

$$q = \sqrt{q} \quad [q = \sqrt{q}] \quad \& \quad \begin{cases} p = 1 - q \\ p = 1 - \sqrt{q} \end{cases}$$

(ii) To know the fr. of carrier :-

If the HW-eqm. can be assume the fr. of heterozygotes among all the individual will be $2q(1-q)$.

Fr. of carrier among normal individuals ($\frac{p^2}{2pq}$)

$$\frac{pq}{p^2 + 2pq} = \frac{pq}{p(p+2q)} = \frac{q}{(p+2q)} \quad \begin{matrix} \text{recd} \\ \text{F1 exp} \end{matrix}$$

(iii) Test of HW-eqm.

If the data are available for a locus where all genotypes are recognizable the observed fr. of a genotype can be tested for aggregated for a population in HW eqm.

If a population is in eqm. the gene fr. will be same in parents and progeny for this we use χ^2 -test.

$$\checkmark [H^2 = 4pq] \rightarrow \text{for testing HW-eqm.}$$

Gene fr. of multiple allele

In ABO blood group system, there are 4 types of blood groups A, B, AB, O. These blood groups are controlled by 3 types of alleles I_A , I_B , i . I_A & I_B are co-dominant to each other and i allele is recessive to both alleles.

If the frequency are -

$$I_A = p, \quad I_B = q, \quad i = x$$

$$\text{then } p + q + x = 1$$

$$x = \sqrt{0}$$

$$AB = 2pq$$

$$A = p^2 + 2px$$

$$B = q^2 + 2qx$$

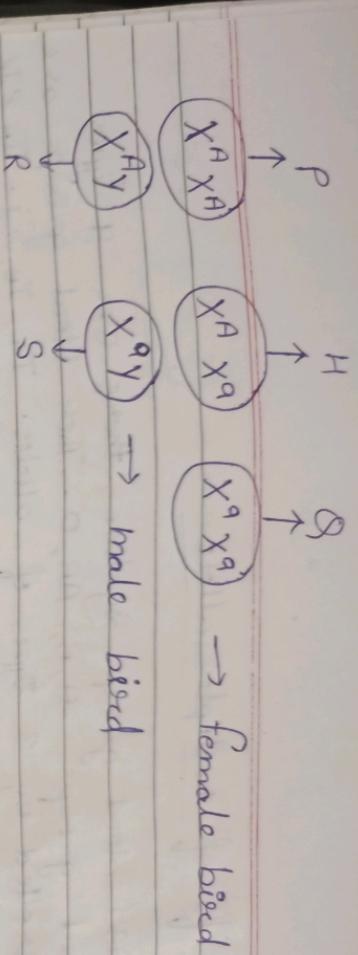
Sex-linked gene :- The relationship b/w the gene fr. and genotype fr. for sex linked gene are diff. than the autosomal genes.

However this relationship is same as with autosomal genes in the homogametic cond? but the heterogametic sex has only 2 genotypes and each individual carries only one gene instead of 2.

For this reason $\frac{2}{3}$ rd of the sex-linked gene in the population are carried by homogametic sex and $\frac{1}{3}$ rd by heterogametic sex.

$$A = p \quad p$$

$$a = q$$



f_r of A in female

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

f_r of a in female

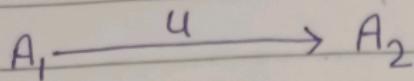
$$f_f = Q + \frac{1}{2}H$$

f_r of A in male [P_m = R]

f_r of a in male [P_m = S]

$$\begin{aligned}
 P &= \frac{2}{3}P_f + \frac{1}{3}P_m \\
 &= \frac{1}{3}(2P_f + P_m) \simeq \frac{1}{3}(2P + H + R)
 \end{aligned}$$

Recurrent mutation - Unidirectional



u = Proportion of A_1 allele which converts into A_2 allele in each generation

Degree of dominance

- Differences of fitness means selection is working
- Here, dominance means dominance with respect to fitness.

No dominance :- Fitness of heterozygote is mean of fitness of both homozygotes.

Partial dominance :- Fitness of heterozygote is b/w mid point and dominant homozygote.

Complete dominance :- Fitness of heterozygote is equal to fitness of dominant homozygote.

Case of selection against a recessive when elimination of unwanted homozygote is complete ($s=1$)

$$q_1 = \frac{q}{1+q}$$

After the t generation -

$$q_t = \frac{q_0}{1+q_0}$$

Generation required to change gene fr. from

$$q_0 \text{ to } q_t = \frac{1}{q_t} - \frac{1}{q_0}$$

When gene fr. is changed by selection some individual must suffer genetic death by their failure to survive or to reproduce & the avg. fitness of the population is reduced.

The proportion of population that suffers genetic death is k/a load.

If L is the load the avg. fitness of population is $(1-L)$ for a recessive gene the avg. fitness is $(1-sq^2)$ and the load is sq^2 .

Dispersive forces :- (Ap)

The dispersive forces are random in direction and predictable only in amount.

In a small population, gene frs. are subject to random fluctuation arising from the sampling of gametes. The gametes that transmitt gene to next generation carry a sample of gene in parent generation and if sample is not large the gene frs. are liable to change b/w one gen. and the next. This random change is of gene fr. is dispersive force.

4 Consequences of dispersive force -

Genetic

① Random drift / Genetic drift

The random changes of gene fr. in a small population in an erratic manner from gen- to generation with no tendency to revert its original value; is k/a genetic drift.

② Differentiation b/w some subpopulation

Random drift occurring independently in diff. sub population leads to dif^{genetic} differentiation b/w sub population.

③ Uniformity within sub population

Genetic variation within each sub population is reduced and individuals become more and more alike within sub in genotype.

This genetic uniformity is the reason for the widespread use of inbred strains of lab. animals. In many areas of biological research

④ Increased homozygosity

Homozygotes increase in fr. have the expense of heterozygote.

In order to reduce the dispersive forces, an idealised population as follows-

Initially one large population in which mating is random and this population becomes subdivided into a large number of subpopulation.

The initial random mating population \rightarrow Base pop and sub-populations \rightarrow line lines.

All the lines together constitute the whole pop. and each line is a small pop. in which gene fr. are subject to the dispersive forces.

The condⁿ specified for idealised pop. are -

- (i) Mating is restricted to members of same line
The lines are isolated in the sense where no genes can pass from line to another; migration is excluded.
- (ii) Generations are distinct and don't overlap.
- (iii) Number of breeding individuals in each line is the same for all the lines and in all gen.
- (iv) Within each line mating is random.
- (v) No selection at any stage
- (vi) Mutation is disregarded.

$$\Delta q = \frac{pq}{2N}$$

N = number of individuals in a population

The change of gene fr. resulting from sampling can be predicted in terms of variance that is $\sigma^2 = \frac{pq}{2N}$

Due to dispersive forces, gene fr. attains the limits 0 and 1

If a gene is lost, then its fr. is 0 & when a particular allele is reached then its fr. is 1 and it is fixed in population.

Effective Population Size

Effective Population Size (Ne) :- Number of individuals that would give rise to calculated sampling variance or rate of inbreeding if they breed in the manner of the idealised population.

To implement the variance in gene fr.

- To evaluate the dispersive forces in real population it must be changed ~~into~~ ^{on} idealised pop.
- The effective pop. size provides a mean to apply the concept of dispersive forces in real population.

Different number of males & Qs \div

The effective population size is twice the harmonic mean of the number of two sexes.

$$\checkmark Ne = \frac{4 N_m N_f}{N_m + N_f}$$

N_m = number of males

N_f = number of females

→ The rate of inbreeding depends chiefly on the number of the less numerous sex.

Values and Mean

Value :- The ~~mag~~ amount of metric unit by which the character is measured.

Phenotypic Value :- The value observed or recorded when the character is measured on an individual is called P.V. of that individual for that character.

P.V. is the joint product of genotype and environment
it means $P = G + E$

(Statement was given by "Johnson")

P = Phenotypic value

G = Genotypic value

E = Environmental deviation

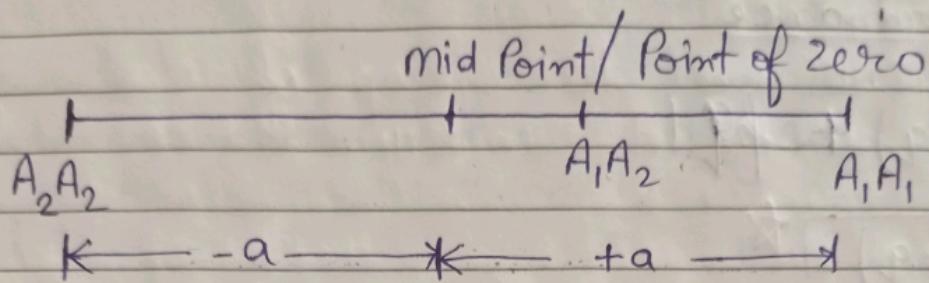
The genotype is the particular assemblage of gene possessed by individual and the environment is all the non-genetic circumstances that influence the P.V. (Housing, Feeding, Behaviour)

The components of value associated with genotype and environment are the genotypic value, phenotypic value and environmental deviation.

Thus the environment causes a deviation in genotypic value before its expression into the phenotypic value.

Population mean :- Considering a single locus A with alleles A_1 & A_2 . Genotypic value of one homozygote is $\oplus q$ and that of other homozygote is $\ominus q$ and for heterozygote is q .

We must have a scale of genotypic value as follows-



Mid point / Point of zero value is the mean of phenotypic values of both homozygotes.

$$\text{Mid point} = \frac{P_{11} + P_{22}}{2}$$

If genotypic fr. of 3 genotypes is p^2 , $2pq$ & q^2
then population mean -

Genotype	Genotypic value (X)	Genotypic fr. (f)	fx
A_1A_1	$+a$	p^2	p^2a
A_1A_2	0	$2pq$	$2pqd$
A_2A_2	$-a$	q^2	$-q^2a$

$$\sum f = 1 \quad \sum fx = p^2a + 2pqd - q^2a$$

$$\bar{x} = \sum fx \quad \sum fx = p^2a + 2pqd - q^2a$$

$$M = a(p^2 - q^2) + 2pqd$$

$$M = a(p+q)(p-q) + 2pqd \Rightarrow M = a(p-q) + 2pqd$$

This population mean is the deviation from mid point.

① No dominance

$$d = 0$$

$$M = a(p-q) + 2pqd$$

$$\boxed{M = a(p-q)} \quad \star$$

$$\boxed{M = a(1-2q)}$$

② Complete dominance

$$d = +a$$

$$M = a(p-q) + 2pqd$$

$$\boxed{M = a - 2q^2a} \quad \star$$

$$= a(1-2q^2)$$

The mean environmental deviation in pop. has a sole is taken to be zero, so that mean phenotypic value is equal to the mean genotypic value.

$$P = G + E \rightarrow \text{zero}$$

$$P = G$$

* The population mean has a deviation is the weighted avg. of genotypic values which is obtained as sum of products of values of each genotype with its f_2 .

$$\boxed{M(\text{Population mean}) = a(p-q) + 2pqd}$$

Average effect :- In a random mating population the average effect of a particular gene/allele is the mean deviation from population mean of individuals, which received that gene from one parent; The gene received from other parent having come at random from population.

(D8)

Average effect of a gene is the mean deviation from population mean of all the genotypes into which this gene enters.

$$A_1 \text{ allele} \rightarrow \alpha_1 = qx[a + d(q-p)]$$

$$A_2 \text{ allele} \rightarrow \alpha_2 = -px[a + d(q-p)]$$

Average effect of (α): When only 2 alleles at a locus are under consideration
Gene Substitution → the average effect of gene substitution is the difference b/w the average effect of 2 alleles and it is denoted by α .

$$\alpha = a + d(q-p) \quad \begin{cases} \alpha_1 = +q\alpha \\ \alpha_2 = -p\alpha \end{cases}$$

Average effect of Substituting one allele for another at random in the population without regard to population mean is the average effect of gene substitution.

$\sigma^2 \rightarrow$ Sire

$\sigma^2 \rightarrow$ Dam

→ Linear regression of genotypic value on number of gene Is the average effect of gene substitution

→ Avg. effects are the prop. of as well as gene Concern.

Important

Breeding Value :-

① The value of an individual judged by the mean value of its progeny.

(08)

② Theoretically, Breeding value of an individual is equal to the sum of avg. effects of all gene it carries. summation being made over the pair of allele at each locus or over all the side.

(08)

③ Practically, if an individual is mated to a number of individual taken at random from population then its Breeding Value twice the mean deviation of progeny from population mean.

Mean deviation of a progeny of an individual from population mean is k/a transmitting ability.

$$\boxed{\text{Breeding value} = 2 \times \text{transmitting ability}}$$

Avg. effect can't be measured but breeding value can be measured.

$$A_1 A_1 \Rightarrow qd + qd = 2qd$$

$$A_2 A_2 \Rightarrow -pd - pd = -2pd$$

$$A_1 A_2 \Rightarrow (q-p)d$$

Dominance deviation variation :- When a single locus is under consideration then the difference b/w genotypic value (G) and breeding value (A) of a particular genotype is k/a dominance variation.

$$D = G - A$$

(I) Interaction deviation :- When genotype refers to more than one locus the genotypic value may contain an additional deviation due to non-additive combination it is k/a Epistatic / Interaction deviation

$$G = A + D + I$$

* Breeding value is the prop. of individual and
✓ of the pop. from which mates are drawn

Variance :- Amount of variance is measured and expressed as a variance, when values are expressed as deviation from population mean, the variance is mean of the squared values. Thus Genotypic variance is the variance of Genotypic Value and Phenotypic Variance is the variance of phenotypic value.

and Environment Variance is the variance of environmental deviation.

$$\begin{cases} P = G + E \\ V_p = V_G + V_E \end{cases} \quad \begin{cases} V_p = V_A + V_D + V_I + V_E \\ \therefore V_G = V_A + V_D + V_I \end{cases}$$

V_A = Additive variance (variance of breeding value)
 V_D = Dominance variance (variance of dominance deviation)
 V_I = Interaction variance

2 Condⁿ must be fulfilled that (i) there is no correlation b/w genotype and environment

(ii) There is no interaction b/w genotype and environment

V_A is the important component which is the chief cause of resemblance b/w relatives and it is the only component that can be readily estimated from observations made on population.

In practice, therefore the imp. partition if into additive genetic variance v/s all the rest (non-additive genetic variance and environmental variance).

This partitioning leads yields the ratio V_A/V_P which is the heritability of character.

$$V_P = V_A + V_{NA} + V_E$$

V_A is obtained by the squaring the values in table multiplying by the fr. of genotype concern and summing of the 3 genotypes.

$$\begin{aligned} A_1A_1 &= 2qd, & 4q^2d^2 \times p^2 \\ A_1A_2 &= (q-p)d, & (q-p)^2d^2 \times 2pq \\ A_2A_2 &= -2pd, & 4p^2d^2 \times q^2 \end{aligned}$$

$$\checkmark [V_A = 2pqd^2]$$

$$\checkmark [V_D = (2pqd)^2]$$

$$\because d = a + d(q-p)$$

$\Rightarrow \therefore$ In case of no dominance

$$\therefore d = 0$$

$$\therefore d = a$$

$$V_A = 2pqd^2 \quad V_D = 0$$

→ Complete dominance

$$(d=1)$$

$$[V_A = \alpha pq \cdot 8pq^3a^2]$$

→ Any degree of dominance -

$$[p = q = 0.5]$$

$$\alpha = a + d(q-p) \\ (\alpha = q)$$

$$V_A = 2pq\alpha^2 \\ = 2 \times \frac{1}{2} \times \frac{1}{2} a^2 \\ = \frac{1}{2} a^2$$

$$V_D = \frac{d^2}{4}$$

→ In case of no dominance, the V_A will be max. when $p = q = 0.5$. and V_D will be zero therefore $(V_G = V_A)$.

→ In case of complete dominance, V_A will be max. when freq. of recessive allele is 0.75 the V_D will be max. when $p = q = 0.5$ and V_G will be max. when $q^2 = 0.5$ or $q = 0.71$

Genotype Environment Correlation

Genotypic value and Environmental deviations are independent to each other and there is no correlation b/w Genotypic Value and Environment deviation but in practical this assumption is Seldom fulfilled

→ Correlation b/w genotype and e is Seldom

→ When better genotypes are given better environment it arises a correlation b/w genotypes and environment and k/a genotype - environment correlation.

e.g. The normal practice of dairy industry is to feed the cattle according to their yield.

When a correlation is present b/w g value and environmental deviation, the phenotypic variance is increased by twice the covariance of genotypic values and environmental deviation.

$$V_p = V_G + V_E + 2 \text{Cov}(G, E)$$

The Covariance is best regarded as part of genetic variance b/c the non-random aspect of environment are a consequence of genotypic value so an individual

environment can be a thought of as part of its genotype

Genotype - Environment Interaction

Assumption

Genotype and environment are additive in nature means there is no interaction b/w genotype and environment and that's why a specific diff. of environment has the same effect on different genotypes.

but practically a specific difference of environment may have a greater effect on genotype than that of others.

Genotype A, may be superior to Genotype B in envir. X but inferior in environment Y.

When interaction b/w genotype and environment is present, the phenotypic value of an individual also has an interaction component (I_{GE})

The interaction component give rise to an additional source of variation and now the equation is -

$$\checkmark V_p = V_G + V_E + 2 \text{Cov}_{GE} + V_{GE}$$

The variance due to genotype environment interaction has being part of the environmental variance

* Different genotype respond differently in different environment, k/a genotype - environment interaction.

G

Phenotype - Environment Interaction becomes very imp. if individuals of a particular pop. are to be reared under different cond.
eg. A breed of livestock may be used by diff. farmers who treat it differently

* Reduction in breeding value, when population perform in environment other than that it was selected is k/a genetic slippage

→ If there is no interaction the best genotype in one environment will be the best in all.

but if there is much interaction the particular genotype must be sort for particular environment

Environmental-Variance

It is a source of error that reduces precision in genetic studies and the aim of experimenter/breeder is therefore to reduce it as much as possible by careful management or proper design of experiment.

Nutritional and climatic factors are the commonest external causes of environmental variation.

Maternal effects are another source of environmental variation particularly in mammals apart from this error of measurement is also an important source of variation. These variations can be minimised by accurate measurement.

In addition to variation arising from recognisable sources there is usually a substantial amount of non-genetic variation where cause is unknown and which can't be eliminated by experimental desire this is genotype.

Resemblence b/w Relatives

Previously, the phenotypic variance was partitioned into components attributable to different causes. These components are called as causal Component of variance (V).

Phenotypic variance can be partitioned into components corresponding to the grouping of individuals into families. These components can be estimated directly from phenotypic value and for this reason they are k/a observational component of phenotypic variance (σ^2)

By analysis of variance, the total observed variance is partitioned into 2 Components Between groups & Within group.

Between group Component is the variance of the true means of group about population

Within group Component is the variance of individual about the true mean of their group

* Resemblance is expressed as the -
Between G as a proportion of the total Variance, this is the interclass correlation Coffecient (f)

$$\sigma_p^2 = \sigma_B^2 + \sigma_W^2$$
$$f = \sigma_B^2 / \sigma_p^2 \text{ or } \sigma_B^2 / (\sigma_B^2 + \sigma_W^2)$$

$\tau_B^2 \rightarrow$ Between Group Component
 $\tau_W^2 \rightarrow$ Within Group Component

BGC (τ_B^2) expresses, the amount of variations that is common to members of same group and aka covariance of the members of the group.

In case of resemblance b/w
 Page 158 Important ★★

Relatives

Covariance

Regression/
 (b/t) Correl

① Offspring & One parent	$\frac{1}{2} V_A$	$b = \frac{1}{op^2} \frac{VA}{V_P}$
② Offspring & mid parent	$\frac{1}{2} V_A$	$b = \frac{1}{op} \frac{VA}{V_P}$
③ Half siblings	$\frac{1}{4} V_A$	* Common environment
④ Full siblings	$\frac{1}{2} V_A + \frac{1}{4} V_D + V_{EC}$	** Variance

$$③ * t = \frac{1}{4} \frac{VA}{VP}$$

$$④ ** t = \frac{1}{2} VA + \frac{1}{4} V_D + V_{EC}$$

$\overbrace{\qquad\qquad\qquad}^{VP}$

Heritability :- Proportion of variations of a trait within population that can be attributed to genetic factors.

Proportion of population that suffers genetic death is k/a load.