POPULATION GENETICS

- Hardy Weinberg law
- Eugenics
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- Inbreeding increases homozygosity

Population Genetics

Population may be defined as a group of interbreeding individuals of a species, living in a particular area at a particular time. The study of inheritance of phenotypic traits in a given population is called *population genetics*. This is founded on a principle proposed independently by *Hardy* in England and *Weinberg* in Germany (1908).

The basic principles of population genetics can be derived directly from Mendelian inheritance.

Organic evolution and the diversification of plant and animal species are to be understood only through an understanding of the population genetics. A population of a particular species includes many inbreeding groups. The inbreeding groups within a particular area are called *Mendelian populations* or *Genetic populations*. A genetic population thus is a group of sexually reproducing diploid organisms with genetic relationship residing in a particular area where interbreeding occurs.

Gene Pool and Gene Frequency

The sum total of genes of all the individuals of a Mendelian population is called **gene pool**. The proportion of a gene to its allele in an interbreeding population is called **gene frequency**. The frequency of a phenotype in a population depends on the frequency of the allele controlling it. So the frequency of a gene in a gene pool of a Mendelian population can be calculated from the observed frequencies of certain phenotypes.

Example: In human population, certain people find bitter taste for the chemical *phenylthio-carbomide* (PTC); These persons are called *tasters*. For other persons it is tasteless. These persons are called *non-tasters*. The bitter taste is controlled by a dominant gene T and its recessive allele in its homozygous condition (tt) produces tastelessness. Thus, genotypically there are three types of human beings namely homozygous tasters (TT), heterozygous tasters (Tt) and homozygous non-tasters (tt).

Assume that there are 100 people in a human population. Out of which, 30 are homozygous tasters (TT); 50 are heterozygous tasters Tt and 20 are homozygous tasters (tt). As each person contains two alleles for the character, the total number of genes in the population is $100 \times 2 = 200$. The total number of dominant genes (T) in the population is 60+50=110. (60 genes in 30 homozygous tasters (TT) and 50 genes in 50 heterozygous tasters (Tt).) Thus the frequency of T in the population is 110/200 = 0.55. Similarly the total number of recessive genes (t) in the population is 40+50=90 (40 genes in 20 homozygous non-tasters (tt) and 50 genes in heterozygous tasters (Tt).) Thus the frequency of t in the population is 90/200=0.45.

The total frequency of T and t in the population is 1. If T is represented by p and t is represented by q, then p+q=1; p=1-q; q=1-p.

Hardy-Weinberg Law

Hardy-Weinberg law was proposed by *Hardy* and *Weinberg* in 1908.

It states that gene frequencies of various genes of a population remain constant generation after generation when,

the population is large,

mating is at random

and in the absence of

mutation, selection and migration.

When the gene frequency remains constant generation after generation, the population is in *genetic equilibrium* or *Hardy* - *Weinberg equilibrium*.

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

$$(\mathbf{p} + \mathbf{q} = 1)$$

p = Frequency of a dominant gene

q = Frequency of a recessive gene

This formula is used to find out the frequency of dominant gene and recessive gene in a population.

The frequency of heterozygotes and homozygotes in a population can be calculated by another Hardy-Weinberg formula:

$$(p+q)^2 = p^2 + 2pq + q^2$$

= 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.

Significance of Hardy-Weinberg Law

- 1. This law states that the gene frequencies in a large population, remain constant generation after generation when mating is at random and when there is no selection and mutation.
 - 2. In small populations, this equilibrium cannot be maintained.
- 3. When the population is in equilibrium there is no possibility for evolutionary change and hence the rate of evolution is zero.
- 4. Evolution occurs only when this equilibrium is upset or altered.
- 5. The equilibrium is detrimental and it prevents evolutionary progress.
- 6. The equilibrium tends to conserve gains (of characters) that have been made in the past and to prevent too rapid changes.
- 7. The equilibrium keeps a store of recessive genes continually in existence in the population.
 - 8. The equilibrium maintains heterozygotes in the population.

Applications of Hardy - Weinberg Law

Hardy-Weinberg law has two main applications:

- 1. Calculation of frequencies of recessive and dominant genes in a population.
 - 2. Calculation of heterozygotes in a population.

Let us assume a hamster population containing 1000 individuals of these 300 individuals are *grey* with recessive genes *mm*. The remaining 700 individuals are *black*. The black individuals may be homozygous dominant *(MM)* or heterozygous *(Mm)*. The two types of black individuals cannot be identified externally.

By applying Hardy-Weinberg formula, the frequency of M and m genes can be calculated. Similarly the frequency of heterozygous individuals can be calculated.

Total population = 1000
Grey (mm) = 300
Black (MM and Mm) = 700
Frequency of Grey =
$$\frac{300}{1000}$$
 = 0.3
Frequency of Black = $\frac{700}{1000}$ = 0.7

Let the frequency of M gene is = p
The frequency of m gene is = q
Apply the Hardy - Weinberg formula

$$(p+q)^2 = p^2 + 2pq + q^2$$

= $0.7^2 + 2 \times 0.7 \times 0.3 + 0.3^2$
= $0.49 + 0.42 + 0.09$
= $0.49 \text{ MM} : 0.42 \text{ Mm} : 09 \text{ mm}$
= $49\% \text{ MM individuals}$
= $42 \% \text{ Mm individuals}$
= $9\% \text{ mm individuals}$

So the frequency of heterozygote in the population is 42%. So out of 1000 individuals 420 are *Mm* individuals, 490 are *MM* individuals and 90 are *mm* individuals.

The frequency of M and m genes can be calculated as follows:

Total M genes
$$= 1400$$

Frequency of M genes (p) $= \frac{1400}{2000}$
 $= 0.7$
90 mm individuals contain $= 180$ m genes
420 Mm individuals contain $= 420$ m genes
Total m genes $= 600$
Frequency of m gene (q) $= \frac{600}{2000}$
 $= 0.3$
 $= 1$
 $= 1$

Eugenics

Eugenics is the science of production of healthy offspring by the application of the laws of genetics to the improvement of the human race. The primary aim of eugenics is to produce a race of physically perfect human beings.

The eugenic measures involve two types and they are *negative eugenics* and *positive eugenics*.

Negative Eugenics

It refers to preventing the multiplication of defective genes. Negative eugenic measures are as follows:

1. Regulation of Marriages

Characters of the offspring, are determined by the parental stock. Number of abnormalities like albinism, colour blindness and some fatal defects like haemophilia are transmitted from parent to offspring. Persons with hereditary diseases should not be allowed to marry. So they are excluded from reproduction and the future progeny is saved from these hereditary defects.

2. Isolating the Defective Persons

Peoples with serious abnormal hereditary defects, like epilepsy, feeble mindedness and criminal tendencies should be isolated. Such persons should not be permitted to mingle and marry with normal persons.

3. Sterilizing the Defectives

The genetically defective persons should be sterilized. The process of sterilization involves *tubectomy* (removal of oviduct) in the female and *vasectomy* in the case of males. The vasectomy is the partial removal of vasa deferentia and preventing the release of spermatozoa.

4. Prevention of Consanguineous Marriages

Consanguineous marriages (cousin marriages) increases the frequency of appearance of recessive defects. The lethal genes are recessive and so remain unexpressed in the heterozygous condition. Marriages between close relative like cousins, tend to bring together

the recessive alleles in homozygous condition. Such close marriages should be restricted.

5. Controlled Immigration

The diseased, feeble minded persons or persons with hereditary defects should not be permitted for immigration.

6. Birth Control

People with genetical defects should be advised to have small family.

Positive Eugenics

It comprises of all those eugenic measures, which help to increase the percentage of desirable traits in the population.

1. Early Marriage

Persons with desirable characters should marry early. They should produce more children. Late marriages produce defective children.

2. Selective Mating

The selected men and women of best eugenic value should be encouraged to increase their birth rate.

3. Selective fusion of Gametes

The sperm and ova from most desirable male and female should be collected and stored in *egg* and *sperm banks*. These selected sperm and ova are used for artificial insemination and implantation and produce test tube babies.

4. Removal of Social Hindrances

Religion, castism and dowry are major social hindrances in the way of marriages. Because of this, good germplasm from different sources fails to come together. These social hindrances should be removed and inter-religion, inter-caste and inter-racial marriages should be encouraged.

5. Education

The people should be given proper education on human biology, human genetics and sex.

6. Gene therapy

In gene therapy, a defective gene is replaced by a normal gene by applying genetic engineering techniques.

7. Genetic Counselling

The advice given for the improvement of hereditary characters is called *genetic counselling*.

The person giving the advice is called **genetic counsellor**. Advice is given on the basic principles of inheritance.

Euthenics

The science of improvement of existing human race is called *euthenics*. It is achieved by the following methods:

1. Pollution Control

The living place should be pollution free. The environment should be neat and clean. Pollution must be controlled at the global level.

2. Balanced Diet

Nutritious food should be given to the children and adults.

3. Selection of Children

Children with superior traits should be selected by applying intelligent tests.

4. Creation of Superior Students

The children must be made into superior, intelligent and talented students. This is achieved by the following methods:

- 1. Admitting the students in standard schools.
- 2. The curriculum (syllabus) must be modernized with latest equipments.
- 3. The students must involve in extracurricular activities like sports, debates, quizes, scouting, military training, cultural activities, etc.
 - 4. The habit must be regularized on good discipline.
 - 5. The students must visit industries for motivation.

5. Medical Facilities

All kinds of medical facilities should be given to the children.

Euphenics

- Euphenics is the study of phenotypic improvement of humans after birth.
 - Euphenics literally means normal appearing.
- Euphenics aims to improve the outcome of a genetic disease by altering the environment.
 - The term 'euphenics' was coined by Lederberg (1963).
- *Phenylketonuria* is a genetical disease is an inborn error in metabolism.

A child born with this disease can avoid the expression of the disease by staying on a low *phenylalanine diet*.

Inbreeding

Mating between closely related individuals is called inbreeding. Self fertilization is an ideal inbreeding.

Mendel carried out inbreeding among the F₁ plants in his *mono-hybrid* and *dihybrid experiments*.

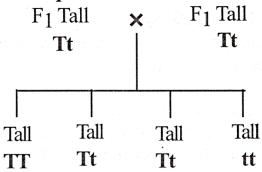


Fig.: Inbreeding in Mendel's experiment.

The marriage between a brother and a sister (not in practice) is an ideal inbreeding.

The Royal family of Egypt including Cleopatra was famous for inbreeding between brothers and sisters.

Cousin marriages are examples of inbreeding.

The mating between a mother cow and an Ox born for the cow is an inbreeding.

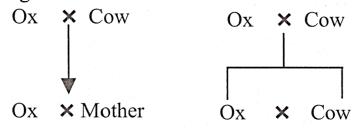


Fig.: Examples of inbreeding.

Significance of Inbreeding

Inbreeding has demerits and merits.

Demerits of Inbreeding

1. Low Yield

Inbreeding results in low yield.

2. Inbreeding Depression

The loss of vigour as a result of inbreeding is called **inbreed**ing depression.

3. Appearance of Deleterious Characters

In human beings, there is a seven-fold increase in phenylketonuric children from cousin marriages than from marriages between unrelated parents.

In corns, inbreeding results in the appearance of deleterious characters such as white seedlings, yellow seedlings and dwarfs.

Merits of Inbreeding

1. Increase of Homozygotes

In an inbreeding population, the homozygotes increase and the heterozygotes decrease. In the course of time, *heterozygotes* are *eliminated* from the population.

Let us assume a population containing 1600 *heterozygous* (Dd) individuals. These individuals mate (inbreed) among themselves and produce offsprings in the proportion of 1*DD*:2*Dd*: 1*dd* or 400*DD*: 800*Dd*: 400*dd*.

In the second generation, there will be 600 **DD**, 400 **Dd**, 600 **dd** and so on.

The heterozygotes will be decreased by half in each generation, with corresponding increase in the frequency of homozygotes.

2. Production of Pure lines

Inbreeding produces homozygotes. The homozygotes reproduce only homozygotes by inbreeding. These homozygotes are pure lines as they breed pure. Hence inbreeding produces pure lines.

3. Elimination of Deleterious Recessive Characters

Inbreeding produces recessive homozygotes. If the recessive character is deleterious, it is possible to eliminate the undesirable recessive genes from the population by repeated inbreeding and selection.

4. Production of Valuable Breeds

High quality commercial breeds of plants and animals are produced by inbreeding followed by selection. Best races of horses, dogs, bulls and sheep are produced by inbreeding and selection. This kind of inbreeding is also called *line breeding*.

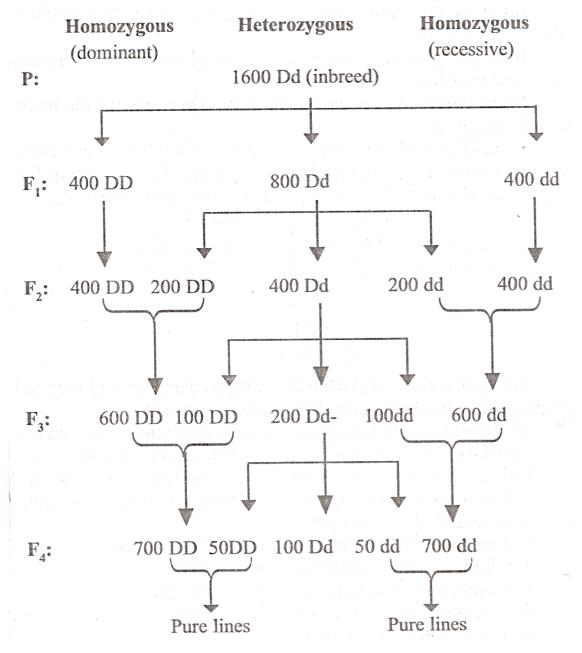


Fig.: Inbreeding