A gene library is a collection of many of the desired genes of DNA fragments maintained in clones of bacterial or some other cells. It is prepared by the following method.

DNA fragments containing one or few desired genes are obtained with the help of specific restriction endonucleases. Each fragment is joined to a suitable vehicle DNA to form recombinant DNAs of different nature. These are then introduced into host (bacterial, yeast, plant or animal) cells. The cells containing recombinant DNAs are allowed to multiply in cultures. This will produce clones of cells where the daughter cells carry the same genes which are identical to those of parent cells. A collection of clones with recombinant DNA containing desired genes is a gene library. Gene libraries are maintained through special techniques.

A gene bank is a store house of clones of known DNA fragments, genes, gene maps, seeds, spores, frozen sperms or eggs or embryos. These are stored for possible use in genetic engineering and breeding experiments where species have become extinct. The need of gene banks is being increasingly felt as the rate of extinction is increasing day by day. The human genome project is the most remarkable contribution in this field.

HUMAN GENETICS

The study (analysis) of genetic characters and aspects like genetic improvements among humans are included in **human genetics.** This is also known as **eugenics** (=well born).

Eugenics is a term derived from Greek language '**Eugenes**' meaning "Well born". First of all, **Sir Francis Galton,** 1883 proposed the idea of improvement in human species through change in hereditary characters in a scientific manner and named it Eugenics. Because of this Sir Francis Galton is known as "father of Eugenics".

To find out various facts, scientists have to perform a number of experiments, but it is not possible to do so in humans. The folowing problems are faced in studying human genetics -

1. Cells of human body are relatively smaller and number of chromosomes present in them is more.
2. It is not possible to perform various experiments on humans in the laboratory.
3. Due to greater life span of humans, a lot of time is required to study their genetic characteristics.
4. Rate of reproduction is slow in humans.
5. Individuals of human species are generally heterozygous for various characters and it is very difficult to find homozygous individuals.
6. Due to controlled hybridization and long lived life among humans, it is not possible to study many generations in easy way.

Despite of above mentioned problems humans are considered suitable for genetical experiments due to-

1. Longer life span of humans, the abnormalities that require relatively long period to express themselves can be easily studied.
2. By pedigree study and analysis of families, many genetic characters of man can be traced out.
3. To study many diseases (like haemophilia, colour blindness etc.) and intelligence quotient (I.Q.) humans are more preferable than any other organisms.

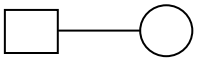
Devices Used in Human Genetical Studies

The study and analysis of human genetics is performed by many methods like pedigree analysis, statistical analysis and human karyotyping. Of these the important ones, that is, pedigree analysis and human karyotype is being described here.

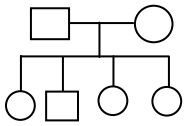
1. Pedigree Analysis

Study of ancestoral history of man of transmission of genetic characters from one generation to next, is pedigree analysis. Dwarfism, albinism, colour blindness, haemophilia etc. are genetically transmitted characters. To study and analyse them a pedigree of genetic facts/data and following symbols are used.

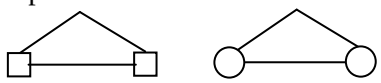
Symbol used in Pedigree :

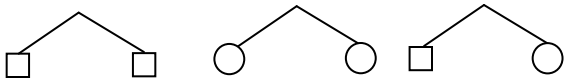
1.  - Normal Male
2.  - Normal Female
3.  - Mating (marriage)

Usual practice is to place the males first, on the left if one parent is not shown in a pedigree, it indicates that this individual was phenotypic ally normal.

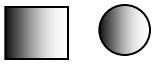
4  The siblings are indicated in chronological order of birth

5.  Sex unspecified

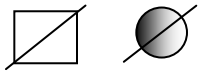
6. Twin
If monozygotic 

If dizygotic 

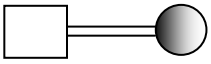
7.  Affected male and female individual

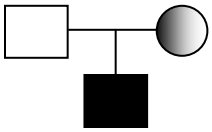
8.  Heterozygous for autosomal recessive

9.  Carrier female of sex linked recessive character or disease

10.  Death of individual

11.  Abortion or still birth (sex unspecified)

12.  Consanguineous marriage

13.  Parent with male child affected with disease

14. 

- Five unaffected offsprings

Pedigree analysis provides valuable informations regarding genetical make up of human beings. If any genetic disease is occuring in a family, then pedigree analysis provides guidance to forthcoming parents about their future progenies for example- polydactyly in humans.

2 Human Karyotype

Humans have 23 pairs (46) chromosomes. In this method, the chromosomes (autosomes and sex chromosomes) | are arranged according to their size and structure. Based on the position of centromere and relative lengths of |

both arms of chromosome, three types of chromosomes are found in human - metacentric, submetacentric and acrocentire. Karyotype helps to know the relative structures (morphology) of chromosomes. Besides, it helps in chromosomal identification and it's nomenclature. It is also used in studying chromosomal abnormalities.

Types of Eugenics

For improvement of hereditary characters in future generations there should be an increase in number of best individuals and reduction in number of defective individuals. Presently, eugenics *is* studied under following types -

1. **Positive Eugenics**

In positive eugenics the main approach is to increase the number of progenies having best hereditary characters. This is achieved through genetic counselling, selective mating (planned marriages), arranging marriages at right age, freedom from social bonds etc.

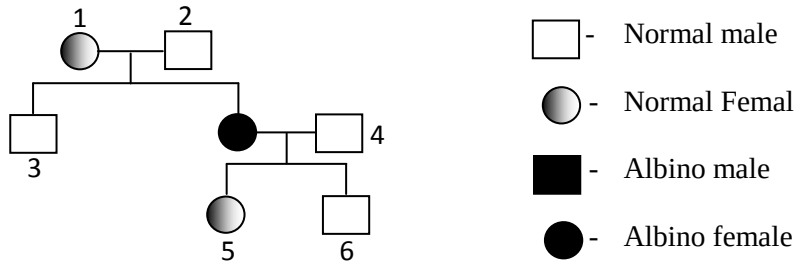
Preservation of ovum of superior quality, artifical insemination and preserving the best quality sperms with aid of modern techniques are also included in positive eugenics.

Euphenics and gene engineering techniques are also being used to establish the best quality of hereditary traits in human society.

2. **Negative Eugenics**

In negative eugenics the carriers of undesirable inferior traits (or characters or genes) are not allowed to produce their progenies by various checks such as marriage control, segregation, birth control, sterilization and control of consanguineous marriages.

Example : Given pedigree shows inheritance of albinism which is an autosumal recessive disorder. If 4th individual is homozygous normal then find out the carrier individual.



POPULATION GENETICS

Study of gene frequency in a population is called population genetics.

1. **Gene pool** - A gene pool is the sum total of genes in reproductive gametes of a population.
2. **Gene-flow** - Migration of gene from one population to another population by cross fertilization.
3. **Genetic load** - The existance within the population of disadvantegeous allele in heterozygous genotype is known as genetic load
4. **Gene frequency** - Gene frequency is defined as proportion of different alleles of a gene in a population.

Ex. In a population of 100 individuals of MN blood group 50 MM, 20 MN, 30 NN find out the frequency of M&N.

MM - 50
MN - 20
NN - 30

TotalMgene- 50x2 + 20 = 120
Total N gene- 30x2 + 0 = 80
Total gene - 200

Freq. of M gene $p\frac{M}{M+N}=\frac{120}{200}=0.6$

Freq. of N gene $\frac{M}{M+N} = \frac{80}{200} = 0.4$

0.6 + 0.4 = 1

p + q = 1

Hardy Weinberg Law -

1908 G.H.Hardy (English methematician) & German Physician, W.Weinbergh independently discovered that an equilibrium is established between frequencies of allele in random mating population and these gene frequency remain constant from generation to generation. This law applicable when factors like mutation, selection & migration, are absent.

Hardy Weinberg theorem or Hardy weinbergh law

$p^2 + 2pq + q^2 = 1$ (A -> p, a —> q, p+q = 1)

AA Aa aa

In this equation frequency of A $\rightarrow$P or the Frequency of Homozygous dominant will be AA-P^2

In this equation frequency of a $\rightarrow$q the frequency of homozygous recessive - q^2

the frequency of Aa = 2pq

The frequency of different genotype produced due to random mating will depend upon the gene frequency and

equilibrium is stablished after one single generation of random mating.

Factors affecting gene frequency :

(1) Mutation (2) Selection (3) Migration (4) Randon drift

Directional-mutation, selection, migration

Nondirectional = random drift

Nondirctional which can not be predicted from one generation to the other.

1. **Mutation** : Mutation lead to introduction of new gene leading to genetic difference. Change in gene frequency due to mutation, will depend upon mutation rate.
2. **Selection (Natural selection)** : By the natural selection there is increase in frequency that are more fit to nature. Selections are associated with superior fitness will increase in frequency in the population.
3. **Migration** : Migration recessive or dominant allele can be introduced into a population from a nearby population. This causes difference in frequency between the two population.
4. **Random drift or Genetic drift.:** Allele frequency can change randomly, sometime increasing and sometime decreasing such deviation in gene frequency will be due to **sampling error.** Due to sampling error, random changes in gene frequencies are called random drif

$$\sigma\sqrt{\frac{pq}{N}}$$

σ - sampling error

p & q - initial gene frequency

N - No. of genes which are sampled

population ↑ sampling error ↓

population↓ sampling error ↑

Q. In a random population frequency of recessive phenotype is 0.09. What is the frequency of heterozygous genotype?

Sol. q^2 - 0.09 '

q = 0.3

p = 1 – q

1 - 0.3 $\Rightarrow$ 0.7

Frequency of heterozygote = 2pq = 2 x 0.7 x 0.3 = 0.42 = 42%

AFLP (Amplified fragment length polymorphisms)

This procedure was described by scientist Zabeau & Vos (1993). In this procedure DNA is cut by restriction *enzyme* then these restricted fragment are amplified by P.C.R. and band pattern of these restricted fragments is visualised after gel electrophoresis.

Procedure (3 steps)

(1) Digestion of total cellular DNA with one or more restriction enzymes.
(2) Selective amplication of some of these fragments by P.C.R. primer. .
(3) Electrophoretic separation on gel matrix followed by visualisation of band pattern of these restricted fragment.

R.A.P.D. (Random amplification of polymorphic DNA)

This is a type of P.C.R. in which random (unknown) DNA fragments are amplified.

Bioinformatics

Definition : Bioinformatics is application of computer technology to the management of biological information. Computer techniques are used together, store, analyze and integrate biological and genetic information which can be applied to gene based drug discovery and development.

Drug design based on bioinformatics

Bioinformatics is a new approach for drug designing. In this procedure all the knowledge about the disease is collected, analysed and find out a 'target molecules' that cause the disease. Structure of these molecules is analysed by X-ray or N.M.R. technique, then drugs are developed which can bind and block the activity of these molecules.

Biological data base

Biological data base is collection of genomic, proteomic and metabolic data assosiated with computer software, which
include nucleotide sequence of gene or amino acid sequence of protein, information about structure, function, location
of genes and protein
Importance -

(1) Easy asses to the information.

(2) A method for extracting only required information.