1

Introduction

One of the most important subdivisions of the science of biology is **genetics**, the study of heredity. The term genetics was first coined by WILLIAM BATESON in 1906. The word genetics is derived from the Greek term **gen** which means 'to become' or 'to grow into'. Thus, the derivation of the word indicates that genetics deals not only with the way in which characteristics are transmitted from one generation to the next, but also with the actions of the units of heredity as they bring about the characterestics which they control. According to WEBSTER, **Genetics is the branch of Biology which deals with heredity and variation among related organisms largely in their evolutionary aspects.** The science of genetics originated in 1900 with the rediscovery of a scientific article originally published in 1866 by GREGOR JOHANN MENDEL. This most important branch of biology had its own historical account. Early philosophers and workers had forwarded various speculations and theories to explain the phenomenon of inheritance.

Vapour theory

The Greek philosopher PYTHAGORAS (600 BC) speculated that a moist vapour liberated from the body of the male is responsible for the development of an embryo in the uterus of the female.

Fluid theory

The Greek philosopher EMPEDOES thought that each parent produces a fluid known as 'semen' which arises directly from the various parts of the body and is responsible for the production of an embryo in the uterus of the female. ARISTOTLE (384-322 BC), the Greek philosopher was of the opinion that the offspring was produced by the mixing of man's and woman's blood during copulation. He regarded man's semen as a highly purified form of his blood which mixed with the woman's fluid. He considered the woman's menstrual fluid as female semen.

Preformation Theory

ANTON VON **LEEUWENHOCK (1677)** thought that sperms

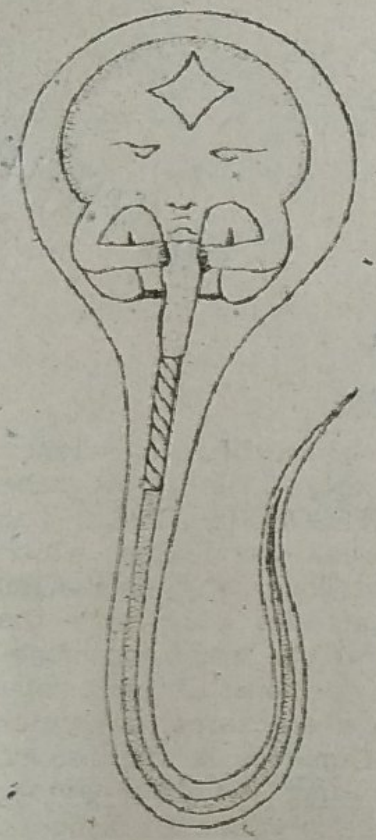


Fig.1: Preformed embryo (Homunculus) in a spermatozoon.

were potential human beings capable of developing into young ones, if introduced into a woman's womb. Another biologist, HARTSOCKER (1695) published a figure showing a tiny (small) human being known as **mankin** or **homunculus** in the head of the human spermatozoa. The idea thus developed that the sperm contained the preformed human body, which required a suitable environment to grow into a child.

According to MALPIGHI (1628-1694) the ovum contains a more or less perfect minature of the adult animal. The development consists of the unfolding and increasing in size of the already formed creatures into the adult form.

Epigenetic theory

This theory was proposed by CASPER FRIEDRICH WOLFF (1759). He found that the egg was formed of only granular substances. These granules after fertilization, arranged themselves to form cellular layers called germinal layers. During development these germinal layers are modified into the body of the embryo. This method of progressive development from the simpler granules to the most complex body was referred to as **epigenesis.** This theory is similar to our present day gene concept of heredity.

Particulate theory

MAUPERTIUS (1698-1759) stated that both the parents produce the semen which contains many tiny particles. During copulation the semen of both parents united and formed the embryo. Each organ of the embryo was supposed to be formed by the fusion of two particles, produced by the parents.

Pangenic theory

This theory was proposed by DARWIN (1838). He assumed that each cell of the body of an organism produced a minute copy of its own called GEMMULES or PANGENES. The gemmules were liberated into the blood and were deposited in the testes and ovaries. Thus the gonads were the store houses for the various types of gemmules. The gemmules were then given to the gametes. The young one formed

from the gametes would be having all the 'gemmules characteristic of the parents.

Germplasm theory

This theory was proposed by AUGUST WEISMANN in 1904. It explains both heredity and development of an animal. According to Weismann, the body of an organism is formed of two types of cells, namely **somatic cells** and **germ cells.** The main bulk of the body is formed of somatic cells. The somatic cells disappear with the death of the animal. So any change affecting these cells is not heritable. The reproductive cells (gametes) are said to be germ cells. The germ cells are carried to the descendants generation after generation. So any change affecting these cells is inherited. Each part of an animal is represented in the sex cells by a separate particle called **determinant.** It is located in the chromosome of the nucleus just as the modern genes. The sum total of determinants would represent the various parts of the adult organism. The complete set of determinants would be handed down from generation to generation, and the transmission of determinants would account for the transmission of hereditary characters.

2

Mendelism

2M

The contribution of MENDEL to Genetics is called **Mendelism.** MENDEL is called the **Father of Genetics.** He was born in a peasant family in 1822 in Austria. In 1843 he entered the monastery at Brunn and in 1847 he was ordained as a priest. After completing the theological studies, he worked as a substitute teacher in Mathematics and Greek. In 1851 he was sent to the University of Vienna, where he studied science. Then he returned and worked as a teacher of Physics and Natural Science. He passed his rest of his life as the Abbot. He died in 1884.

MENDEL was fond of gardening from his boyhood. When he was working as teacher, he performed a series of experiments with pea plants in the garden of the monastery. His work contains inheritance of characters in 22 varieties of garden peas. His papers were published in 1866 and 1867 in the proceedings of Natural History Society of Brunn.

The work of Mendel remained unnoticed to the world for 33 years. In 1900 the principles of Genetics worked by Mendel were rediscovered by three botanists, namely CORRENS, De VRIES and TSHERMARK. The unrecognised papers of Mendel were taken out from the grave and made known to the scientific world. When Mendel's work was recognised and appreciated, he was no more.

Rediscovery of Mendel's Principles

In 1900 Mendel's principles were rediscovered by three different investigators. They were DE VRIES (Holland), CORRENS (Germany) and TSCHERMAK (Austria). After Mendel's death several other experiments on plants and animals were done all over the world and Mendel's conclusions were confirmed. Bateson confirmed Mendel's work by a series of hybridization experiments.

Reasons for Mendel's success

Mendel did his work by collecting several types of garden pea **Pisum sativum** from salesmen and studied the differences among them. Then he did hybridization experiments with different types of pea plants. The secret of Mendel's success lay in his wise selection. The following are the reasons for the success of MENDEL:

1. The flowers of pea plants are normally self-fertilized.
2. The pea plant shows a number of clear-cut contrasting characters.
3. The hybrids of garden pea are perfectly fertile.
4. Cross pollination is not very difficult in pea plant.
5. Artificial fertilization is almost always successful.
6. The genes for the seven pairs of characters are located on seven separate homologous pairs of chromosomes.
7. Many pure breeding varieties are available for the experiments.
8. It is very easy to cultivate the pea plants in open ground.
9. They have a short growth period and a short life cycle.
10. He studied the inheritance of only **one character at a time.** This made the complex problem simple.
11. He maintained **statistical records** of the results. It helped Mendel to derive numerical ratios of significance.

Characters selected by Mendel

The pea plant contains a number of contrasting characters. Out of these contrasting characters Mendel selected only **seven characters.** Each of these seven characters has two **varieties** or **alternatives.** The seven characters and their **contrasting alternatives** are shown on the table below.

Mendel in his first experiment crossed two plants differing in one character (height) only. A plant having a tall stem was crossed with another plant having a dwarf stem. Tall and dwarf are the two varieties (alternatives) of a single character, height. Such **crosses, where parents differ in one** pair of alternative characters are known as **monohybrid crosses.** The resulting hybrids are known as **monohybrids.** When the behaviour of each single character was established, Mendel

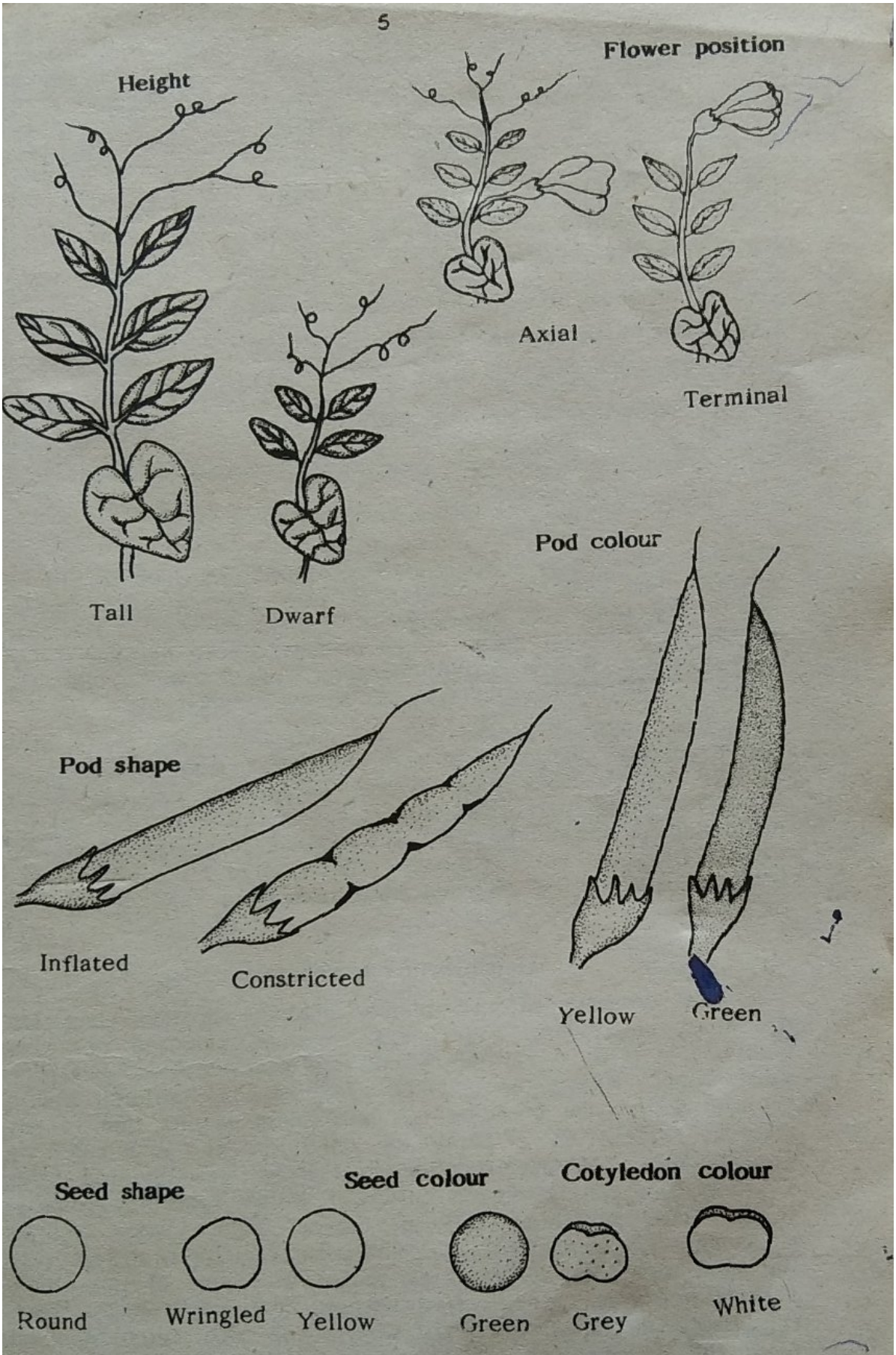


Fig. 2 .Characters selected by Mendel in pea plant.

No.	Characters	Alternatives	
		Dominant	Recessive
1.	The length of the stem	Tall	Dwarf
2.	The position of the flower	Axial	Terminal
3.	The colour of the pod	Green	Yellow
4.	The shape of the pod	Inflated	Constricted
5.	The shape of the seed	Round	Wrinkled
6.	The colour of the seed coat	Coloured	White
7.	The colour of the cotyledon	Yellow	Green

Table showing the characters selected by MENDEL

crossed two plants differing in two characters, such as flower position and height of the stem. A plant having axial flower and a tall stem was crossed with a plant having terminal flower and a short stem. Such **crosses, where parents differ in two pairs of alternative characters are known as dihybrid crosses.** The resulting hybrids are known as **dihybrids.**

The plants involved in the above crosses are called **parent** plants. They form the **parental generation** which is marked by P. The first hybrid generation resulting from a cross between parental plants is called the **first filial generation** and is marked as F1. The second generation of hybrids arising from the self or cross-fertilization of F1 hybrid generation is called the **second filial generation** and is marked as F2.

Monohybrid Experiment

The crossing of two plants differing in one character is called monohybrid experiment. Mendel selected two pea plants, one with a **tall stem** and the other with a **dwarf** or **short stem.** These plants were considered as parental plants (P) and were pure breeding. A **pure plant** is one that breeds true in respect of a particular character for a number of generations. The pure-breeding tall plant was selected from a stock that had produced only tall plants for many generations. The pure-bred tall and dwarf plants were treated as parents and were crossed. This was done by first removing the male parts (anthers) of an immature flower of tall plant. This flower was then covered with a small paper bag. When this flower matured, the pistil was dusted with pollen received from the dwarf plant. Seeds were collected from this plant. These seeds were sown and a group of plants were raised. These plants constituted the **first filial generation** or **F1 gene-**

3

Genic interaction

(Factor hypothesis)

According to Mendel each character is controlled by a pair of factors or genes. But later discoveries prove that in many cases the expression of a single character is controlled by the interaction of more than one pair of genes. This is called **interaction of genes** or **factor hypothesis.** It was proposed by BATESON and PUNNET. This hypothesis states **that certain characters are controlled by the interaction of two or more genes.**

Non allelic and allelic gene interaction

The genic interaction may occur in between **genes** located in the same chromosome or different chromosomes. This type of genic interaction is known as **non allelic gene interactions.** The genic interaction may also occur between the two alleles of a single type of gene. This type of genic interaction is known as **allelic gene interaction.**

Some of the important forms of genic interactions are as follows :-

1.	Complementary factors	Non allelic gene interaction
2.	Supplementary factors	
3.	Inhibiting factors (Epistatis)	
4.	Duplicating factors	
5.	Complete dominance	Allelic gene interaction.
6.	Incomplete dominance	
7.	Co-dominance	
8.	Blending inheritance	
9.	Cumulative genes or Multiple genes or polygenes	
10.	Pleiotropism.	

Complementary Genes (9:7)

Complementary genes may be defined as, **"two or more dominant genes occurring in different loci of the same chromosome or different chromosomes interact with one another to produce a character but neither of them produces that character in the absence of the other".** The action of these independent genes are complementary.

Flower colour in Sweet Pea: BATESON and PUNNET studied the inheritance of flower colour in sweet pea, **Lathyrus odoratus.** There are two varieties of pea plants, one producing **red flower** and the other **white flower.**

P White x White
CCaa ccAA

Gametes: Ca ca

F_1 Cc Aa
Red

F1 Plants are crossed: Red CcAa x Red CcAa

Gametes: CA Ca cA ca CA Ca cA ca

Gametes	CA	Ca	cA	ca
CA	CCAA Red	CCAa Red	CcAA Red	CcAa Red
Ca	CCAa Red	CCaa White	CcAa Red	Ccaa White
cA	CcAA Red	CcAa Red	ccAA White	ccAa White
ca	CcAa Red	Ccaa White	ccAa White	ccaa White

Fig.14 : Inheritance of flower colour in sweet pea.

The red colour of the flower is due to the presence of a pigment called **anthocyanin.** The anthocyanin is produced from a colourless substance called **chromogen** by the action of an enzyme or activator. The chromogen cannot be converted into anthocyanin in the absence of the enzyme. Thus for the production of red colour both chromogen and enzyme should be present in the plant. In the absence of anyone the red colour cannot be produced.

Bateson found that a dominant gene **C** is responsible for the production of **chromogen.** When this gene is recessive **c** the chromogen cannot be produced. Similarly another dominant gene **A** is responsible for the production of the **enzyme** or **activator** which converts the chromogen into anthocyanin. When the gene is recessive **a** the enzyme cannot be produced and thus chromogen cannot be converted into anthocyanin.

Gene C ⟶ Chromogen

Gene A ⟶ Activator or enzyme

Chromogen + Activator ⟶ Anthocyanin (Red)

As the red flower plant contains both chromogen and enzyme it should possess both types of dominant gene. ie., **C** and **A.** The two white flowered varieties are homozygous for any one or both of the recessive genes of the different loci. So the possible genotypes of the white flowered plants are **CCaa, ccAA** and **cc aa.** The possible genotypes of the red coloured plants are **CCAA, CCAa, CcAa** and **CcAA.**

BATESON crossed a homozygous white flowered sweet pea plant (**CCaa**) with another genotypically different white flowered sweet pea plant (**ccAA**). The F1 offsprings have red coloured flowers. This red type was like that of the original wild variety (remote ancestral type). The appearance of a remote ancestral type in a cross between two true breeding varieties is called as **reversion** or **atavism** or **throw backs.** In this process the progeny does not resemble the parents, but it shows the characterestic features of its remote ancestor. When the **F1** red hybrid plants (**CcAa**) are selfed or crossed in **F2** red and white are produced in the ratio 9:7.

Both the non allelic genes **C** and **A** are complementary in nature. In the absence of either one or both of the complementary genes, white flowers are produced. Thus it is shown that in the presence of both the complementary genes **C** and **A,** the red colour is produced in the flowers. The red flower variety is ancestral type. The colourless white varieties arose by mutation occured from **C** to **c** or **A** to **a.**

inhibit the expression of other genes. A gene that inhibits or masks the expression of another gene (non allelic gene) is said to be **epistatic**. The gene that is masked is said to be **hypostatic.** There are two types of epistasis, namely **recessive epistasis** and **dominant epistasis.** If a recessive gene masks the expression of a dominant non allelic gene then it is known as recessive epistasis, while dominant gene, masks the expression of another non allelic dominant gene, is known as dominant epistasis.

1. Recessive Epistasis : In mice agouti coat is common (wild type) and is characterized by brownish grey colour. In some individuals the coat is black in colour and in some other forms the coat is white in colour (albino). The agouti coat colour is controlled by the dominant gene. A (wild type

Parents : Black male **MMaa** x Albino female **mmAA**

Gametes : Ma, mA

F1 generation: **MmAa** Agouti

F1 individuals crossed. Agouti male **MmAa** x Agouti female **MmAa**

Gametes : MA, Ma, mA, ma MA, Ma, mA, ma

Gametes	MA	Ma	mA	ma
MA	**MMAA** Agouti	**MMAa** Agouti	**MmAA** Agouti	**MmAa** Agouti
Ma	**MMAa** Agouti	**MMaa** Black	**MmAa** Agouti	**Mmaa** Black
mA	**MmAA** Agouti	**MmAa** Agouti	**mmAA** Albino	**mmAa** Albino
ma	**MmAa** Agouti	**Mmaa** Black	**mmAa** Albino	**mmaa** Albino

F2 Generation: Agouti:9, Black:3, Albino:4

Fig. 18 : *Inheritance of coat colour in mice.*

gene). The expression of this dominant gene. A is masked or inhibited by a pair of recessive genes **mm.** Thus the individuals with, **AAmm, Aamm and aamm** genotypes produce albino coat. The dominant gene **M** in the absence of **A** gives black mice. The individuals with **aaMM** and **aaMm** genotypes produce black coat colour. If both the dominant genes are present in an individual, then that individual will produce the agouti coat. Thus **AAMM, AaMM, AaMm** and **AAMm** genotypes produce agouti coat. In this case the dominant gene. **A** is considered as the hypostatic gene to the paired recessive epistatic genes **mm.** When black mice are crossed with albino, all the F1 individuals are agouti in nature.

If the F1 individuals are crossed themselves. they produce 9 Agouti, 3 black and 4 albino offsprings due to recessive epistasis.

Recessive epistatic genes are also present in man. The skin colour in human being is determined by various genes. These genes are responsible for pigment production in the body. In some groups of population the expression of these pigment producing genes are **masked or inhibited by a pair of recessive genes.**

P Black Negro father **PPMm** x Black Negro mother **PPMm**

F_1 Black **PPMM** : Black **PPMm** : Albino PP **mm**

P = Pigment producing gene (hypostatic gene)
mm = Recessive inhibiting genes (epistatic genes)

In the above experiments the albino son received the pigment producing gene (P) from his parents. He also received the recessive epistatic genes (**mm**) from his parents. The two recessive epistatic genes received from his parents inhibit the expression of the pigment producing gene. So the individual is white in colour even though he carries dominant gene for pigment production.

2. Duplicate Recessive epistasis: In man normal hearing is due to two different dominant genes namely **A** and **B.** But the recessive alleles (**a** and **b**) of the two dominant genes are epistatic when they are homozygous. The **paired recessive** allele of one dominant gene masks the expression of another dominant gene. Thus a person with the genotype **aaBB** would be a deaf-mute because the genes **aa** are epistatic to the gene **B** for normal hearing. The paired recessive gene **b** is epistatic to the gene **A** which is responsible for normal hearing. This is a case of **duplicate recessive epistasis** where either of two recessive genes may be epistatic to the dominant allele of the other.

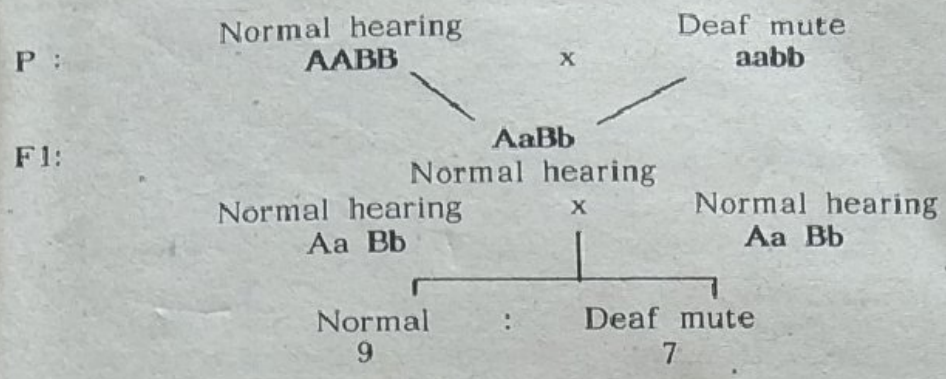
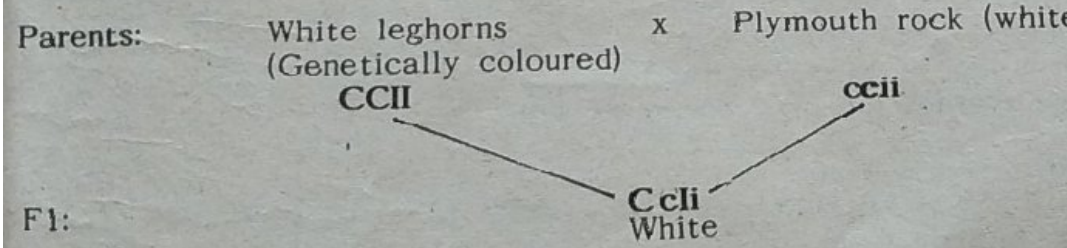


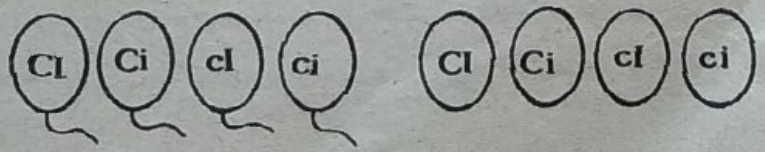
Fig. 19: Inheritance of hearing in man.

3. Dominant epistasis: Out of two genes, the **dominant allele** of one gene masks or suppresses the expression of alleles of another gene. For example, among dogs, the colours of coats depend upon the action of two genes. One gene locus has a dominant epistatic allele **I** and the other locus has a dominant hypostatic allele **C** which is responsible for the pigment production. The inhibitor allele **I** masked the expression of the pigment producing gene **C**.

In poultry there are two varieties of birds that are genetically different. The **white leghorns** contain a gene (**C**) for colour (**hypostatic**) and an epistatic gene (**I**) that inhibits the expression of the colour gene. The genotype of the white leghorn may be **CCII** and that are genetically coloured birds that are unable to develop their colour. In the case of other white varieties Wyandottes or Plymouth rocks there is a factor for white colour (**c**) which is recessive to the factor for colour **C** and the genotype may be ccii. When the white leghorn (**CCII**) and white Plymouth rock (**ccii**) are crossed in the F1 all the offsprings are white. When these F1 varieties are inbred in the F2 generation both white and coloured form appeared in the proportion of about 13 white and 3 coloured forms. These coloured birds appeared in the F2 generations, because the colour factor is free from inhibiting factor.



F1 individuals are inbred — White CcIi x White CcIi



Gametes	CI	Ci	cI	ci
CI	CCII White	CCIi White	CcII White	CcIi White
Ci	CCIi White	CCii Colour	CcIi White	Ccii Colour
cI	CcII White	CcIi White	ccII White	ccIi White
ci	CcIi White	Ccii Colour	ccIi White	ccii White

F2 generation : 13 white. 3 colour

Fig. 20 : Inheritance of colour pattern in poultry.

Biochemical basis of Epistasis: Every cellular chemical reaction involves stepwise conversion of one initial substance called **precursor** to another, called **end product.** All the subsequent steps of a chemical reaction constitute the biosynthetic pathway which includes various steps and each step is catalyzed by a specific enzyme. Each specific enzyme is synthesized by a specific gene. Number of enzymes are involved in the conversion of one material to another material. In other words series of genes are involved in the conversion of one material to another material. For example, the coat colour of an individual is based upon the amount of melanin pigment present in the body. The production of melanin pigment involves stepwise conversion of an initial substance to the end product through a number of metabolites. Each metabolite is produced by the catalytic action of different enzymes which are synthesized by a series of genes.

Initial Substance
↓ ← Enzyme 1 ← Gene 1 **AA** or **Aa**
J-Metabolite
↓ ← Enzyme 2 ← Gene 2 **BB** or **Bb**
K-Metabolite
↓ ← Enzyme 3 ← Gene 3 **CC** or **Cc**
L-Metabolite
↓ ← Enzyme 4 ← Gene 4 **DD** or **Dd**
Melanin (End product)

Fig. 21 : Biosynthetic pathway in normal animals.

A group person has antigen ***A*** and antibody ***b***. B group person has antigen ***B*** and antibody ***a***. AB group person has antigen ***A*** and ***B*** and no antibody. O group person has both antibody ***a*** antibody ***b***.

Table 33.1: Distribution of antigen and antibody in ABO blood group.

Group	Antigen	Antibody
A	A	b
B	B	a
AB	A and B	Nil
O	Nil	a and b

It is thus clear that antigen ***A*** cannot co-exist with antibody ***a*** in any man. Similarly, antigen ***B*** cannot co-exist with antibody ***b***.

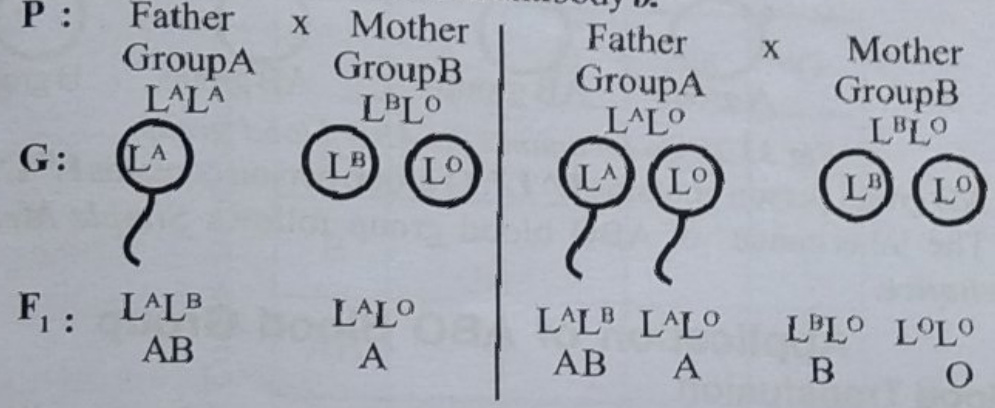


Fig.33.1: Inheritance of ABO blood group.

The synthesis of antigen ***A*** is controlled by a dominant allele represented by L^A (the letter L is used to honour the discoverer of blood groups ***Landsteiner***), Antigen ***B*** synthesis is controlled by another dominant allele represented by L^B. The absence of antigens is due to the presence of recessive allele represented by L^O.

Thus three alleles are responsible for the inheritance of ABO blood group. They are L^A L^B and L^O.

L^O is the recessive allele. It is recessive to both L^A and L^B.

L^A and L^B are dominant alleles. They are ***codominant.*** In codominance, both genes express their character. None is masked. When L^A and L^B occur together in the same man, L^A produces antigen ***A*** and L^B produces antigen ***B***.

As L^A, L^B and L^O occur in the same locus, they are called ***multiple alleles.***

Though there are three alleles, each man contains only two alleles. For example, A group person contains $L^A L^A$ or $L^A L^O$; B group person contains $L^B L^B$ or $L^B L^O$.

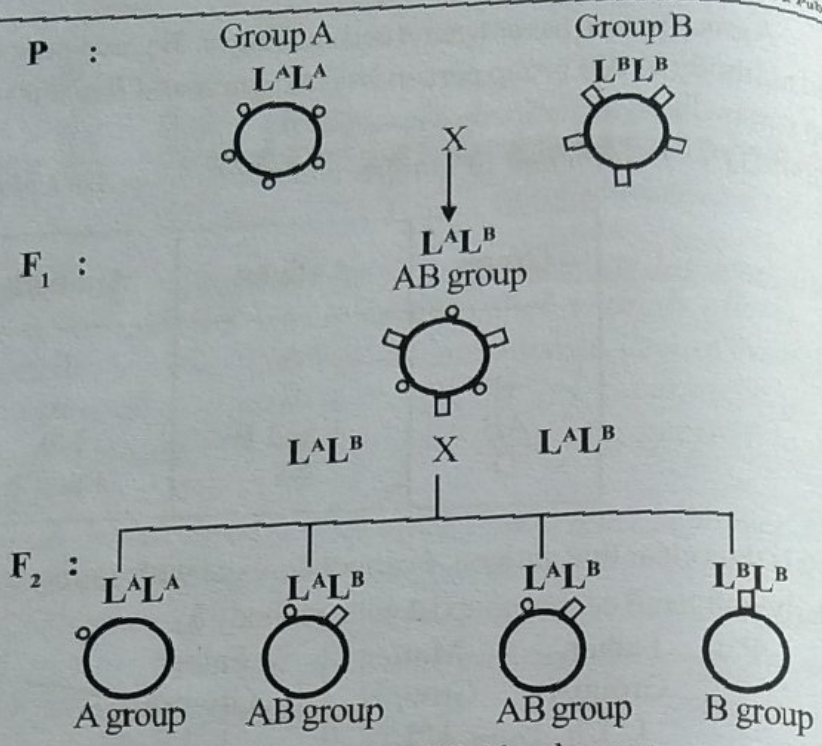


Fig.33.2: Codominance in ABO blood group.

AB group person contains $L^A L^B$; O group person contains $L^o L^o$.

The inheritance of ABO blood group follows ***Simple Mendelian inheritance.***

Application of ABO Blood Group

1. Blood Transfusion

The transfer of blood from one person to another is called ***blood transfusion.*** Blood transfusion is a life saving process in accidents and operation cases.

Loss of blood from the body is compensated by transfusion. Transfusion needs for persons met with an accident or persons undergoing a major surgery or persons affected by anaemia.

Successful blood transfusion needs a thorough blood group testing.

The person donating blood is called ***donor***. The person receiving blood is called ***recipient.*** O group person is the ***universal donor.*** AB group person is called ***universal recipient.***

2. Disputed Parentage

The parents of a disputed baby can be confirmed by testing the blood groups of the doubtful persons.

Similarly, the claim of two mothers for a single child (as it happened in the court of ***King Solomon***) can be settled by blood group testing.

3. Identification of Culprits

In murder cases, the culprits can be confirmed, if the culprit's blood stain is available at the place of murder.

Blood Transfusion

Transfer of blood from one person to another in case of serious loss of blood is termed ***transfusion.***

A person who donates the blood is called the ***donor*** and the person who receives the blood is called the ***recipient.***

In transfusion, if the blood of the donor and recipient are not compatible, agglutination and haemolysis of the donor's corpuscles will take place. While testing the compatibility, the reaction between the antigen of the donor's RBC and the antibodies of the recipient's plasma alone are taken into consideration. The antibodies of the donor has no effect because it is diluted before transfusion.

Plasma of Recipients

Corpuscles of donors	A	B	AB	O
A	-	+	-	+
B	+	-	-	+
AB	+	+	-	+
O	-	-	-	-

+ = Agglutination - = No agglutination

Fig.33.3: Grouping of blood.

The ***AB*** group person is called an ***universal recipient*** because he can receive blood from all groups.

The ***O*** group person is called ***universal donor*** because he can donate his blood to any group.

AB group person can receive blood from all four groups.

O group person can donate blood to any group of persons.

A group person can receive blood from another A group person and O group person.

Similarly B group person, can receive blood from another B group and O group, but not from AB group.

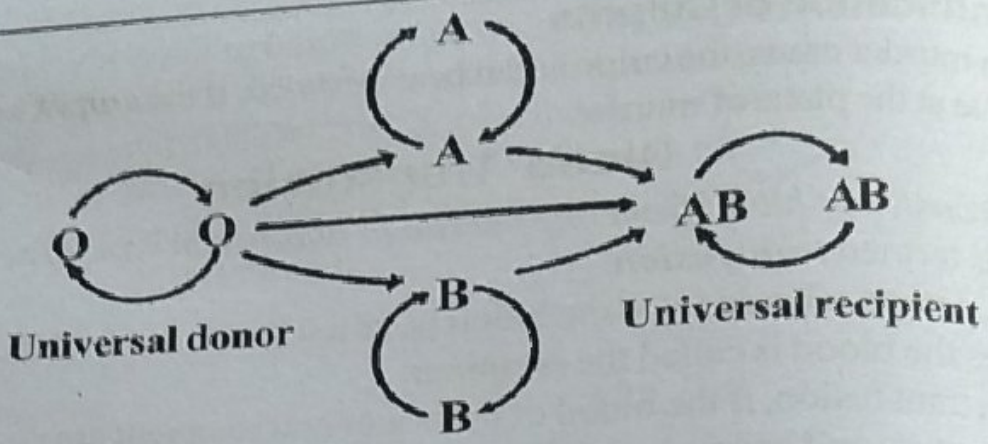


Fig.33.4: Blood transfusion.

Blood transfusion is a life saving process. By blood transfusion, blood can be given to accident cases, major surgery cases and anaemic patients.

34 Rh Blood Group

Rh blood group was discovered by ***Landsteiner*** and ***Wiener*** in 1940.

It is controlled by ***multiple alleles*** located in the same locus of homologous chromosomes.

There are two groups of human beings, namely ***Rh positive*** (Rh+ ve) and ***Rh negative*** (Rh- ve).

Rh positive person contains an antigen called ***Rh antigen*** present on the surface of RBC. Rh antigen is ***Rhesus antigen*** as it was first discovered in ***Rhesus monkey.*** Rhesus antigen is also called ***Rh factor.***

The Rh negative person does not contain Rh antigen.

The Rh antigen has ***no natural antibody.*** However, Rh antibody can be produced artificially. An ***Rh -ve*** person develops Rh antibody when he receives blood from a ***Rh+ ve*** person. Even a small amount of ***Rh+*** ve blood (as small as 0.05ml) can evoke the production of Rh antibody in the ***Rh-*** ve person. The antibody once formed remains in the blood throughout the life.

In the European country, 85% of the human beings are ***Rh+***ve and the remaining 15% are ***Rh-*** ve; in India, 93% are ***Rh+*** ve and 7% are ***Rh-***ve. In China, 99.5% are ***Rh+*** ve and only 0.5% are ***Rh-*** ve

There are several varieties of Rh antigen and of antibody. The commonest Rh antigen is called ***antigen D*** and its antibody is called ***anti D.***

The production of antigen is controlled by multiple alleles. The antigen D is produced by a dominant gene represented by ***R.*** When this gene is recessive, it cannot produce the antigen. Hence the ***Rh***+ ve persons may be homozygous dominant ***(RR)*** or heterozygous ***(Rr).*** The ***Rh***- ve persons are always homozygous recessive ***(rr).***

The ***Rh*** factor follows the ***Mendelian principle of inheritance.*** The blood type of the children can be easily visualized by knowing the blood types of parents.

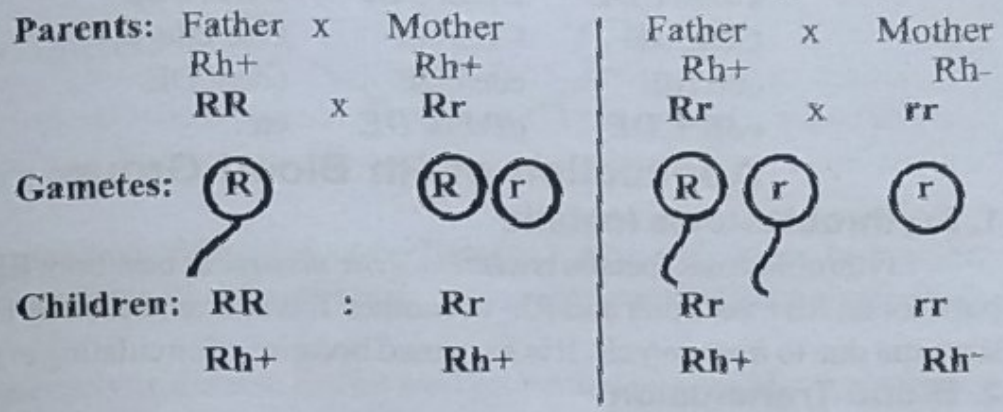


Fig.34.1: Marriages and children of Rh blood group.

There are two views regarding the genes of Rh blood group. They are as follows:

1. Wiener's theory and
2. Fisher's theory

1. Wiener's theory

Wiener proposed that Rh blood group is controlled by 8 multiple alleles, namely

r	R_1
R_o	R_2
R_1	R_x
R_{II}	R_y

Of these ***r*** controls the ***Rh***- ve blood and the remaining genes control ***Rh***+ve blood.

As per this hypothesis, the Rh- ve group is controlled by ***rr*** genes. The genotype of the Rh+ ve group is controlled by any one of the following genotypes:

R_oR_o R_2R_2 R_yR_y R_oR_1 R_1R_x
R_1R_1 R_xR_x R_or R_1r R_1R_y
etc.

2. Fisher's theory

Fisher proposed that three genes are involved in the production of Rh antigens and these three genes are lying so close together on the chromosome that they act as one gene. Such genes are known as ***pseudoalleles.*** He proposed symbols for the three dominant genes as ***CDE*** and the recessive alleles ***cde.*** A Rh negative person should have the genotype ***cde/cde.*** If any one of the dominant gene is present, the person will be ***Rh positive.***

So the different genotypes of the Rh positive persons are as follows:

CDE/CDE	***CDE/CDe***	***CDE/Cde***
CDE/cde	CDe/cde	Cde/cde
cde/cdE	cde/cDE	cde/CDE
cdE/CDE	***cDE/CDE***	etc.

Application of Rh Blood Group

1. Erythroblastosis foetalis

Erythroblastosis foetalis is a ***haemolytic disease*** of new born Rh+ ve babies born for an Rh+ ve father and Rh- ve mother. It is characterized by jaundice and anaemia due to ***haemolysis.*** It is so named because of circulating ***erythroblasts.***

2. Blood Transfusion

The transfer of blood from one person to another needs a blood test analysis of Rh factor.

Erythroblastosis foetalis

Erythroblastosis foetalis is *a haemolytic disease of newborn baby characterized by jaundice and anaemia due to incompatibility of Rh-ve mother and Rh⁺ foetus and haemolysis. It is so named because of circulating* ***erythroblasts.***

Erythroblasts are normally found in the bone marrow. The presence of circulating erythroblasts accounts for the name of the disease.

When an Rh+ ve man marries a Rh- ve woman, the developing baby will be Rh+ ve. The Rh+ ve baby developing in the uterus of Rh- ve mother develops erythroblastosis foetalis.

The Rh antigen of the baby enters the blood of the mother. The blood of the mother produces Rh antibody. When this antibody enters the foetus, the foetus is affected.

The Rh antibody destroys the RBC of the foetus.

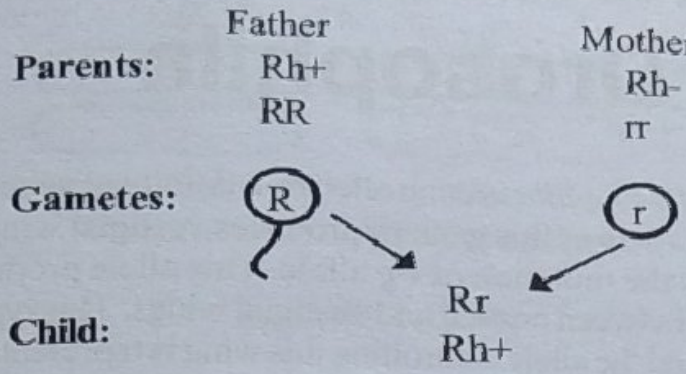
The destruction of RBC leads to haemolytic jaundice and anaemia.

The dead RBCs are carried to liver for disintegration. As more work is given to the liver, the liver is heavily damaged causing jaundice.

This causes the death of the baby.

Prevention of Haemolytic Disease

1. The anti A or anti B prevents the rhesus immunization of the mother. Any Rh+ foetal cells which enter the maternal circulation are destroyed by the mother's naturally occurring anti-A or anti-B before they have time to stimulate the production of Rh antibodies.



2. The destruction of ***Rh***+ ve foetal cells in the maternal blood can be brought about by injection of anti-Rh soon after birth. In this way, maternal Rh immunization can be prevented.

3. The haemolytic disease can be well prevented by selective marriages. When a lady happens to be ***Rh***- ve she should marry only an ***Rh***- ve man and not a ***Rh***+ ve man.

Treatment

The best treatment for severe haemolytic disease is an ***exchange transfusion*** carried out soon after birth. Small quantities of the infant's blood are successively withdrawn and are replaced by an equal volume of compatible ***Rh***- ve blood.

Pseudoalleles

A set of three alleles located very close together controlling one character and inherited together as one block is called ***pseudoalleles.***

The genes controlling Rh blood group are called ***pseudoalleles***. This system was proposed by ***Fisher.***

The Rh antigen production is controlled by a set of three genes that act as one gene. They are ***CDE.*** When any one gene is dominant the person is Rh+ ve. When all the genes are recessive ***cde*** the person is Rh- ve. The genotypes of some persons are as follows:

cde/cde	Rh- ve	CDc/CDe	Rh+ ve
CDE/CDE	Rh+ ve	Cde/CDe	Rh+ ve
CDe/CDE	Rh+ ve	Cde/Cde	Rh+ ve

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1. Agouti

This is the wild type rabbit and its body is ***brownish grey*** in colour. The dominant gene ***C*** is responsible for the brown coat colour. This dominant gene undergoes mutation to give rise to three mutant alleles C^{ch}, C^{h} and ***c*** located in the same locus. These mutant alleles express different shades of coat colours and are recessive to dominant allele ***C***.

2. Chinchilla

In some rabbits, the coat is ***silvery grey*** in colour. The mutant allele C^{ch} is responsible for the production of silvery grey coat colour. This mutant allele is dominant to other mutant alleles C^{h} and c.

Phenotype	***Genotype***
Agouti (wild type)	CC or CC^{ch} or CC^{h} or Cc
Chinchilla (mutant)	$C^{ch}C^{ch}$ or $C^{ch}C^{h}$ or C^{ch} c
Himalayan (mutant)	$C^{h}C^{h}$ or C^{h}c
Albino (mutant)	cc

3. Himalayan

In these individuals, the extremities such as ears, nose, tips of limbs are coloured, while the rest of the body is white. This type of pigmentation is known as ***acromelanism.*** The mutant allele for himalayan is C^{h}. This mutant allele is dominant to the mutant allele ***c.***

4. Albino

From these individuals the pigments, are completely absent. The allele for albino coat is represented as ***c.***

Thus four colour patterns are controlled by a set of four alleles, namely, C, C^{ch}, C^{h} and ***c***. The cross between the four types are given below:

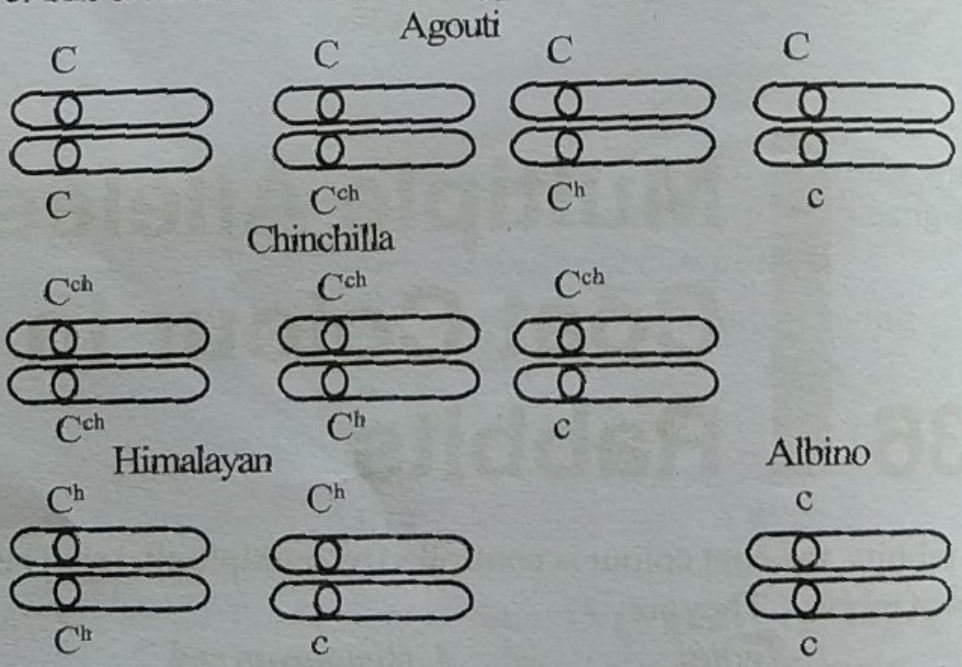

Fig.36.1:Possible genotypes of rabbits with different coat colour.

According to Davenport, skin colour in man is determined by cumulative genes. A pure Negro has two pairs of genes for the presence of pigment in his skin. The genes **P1** and **P2** represent presence of pigment and their recessive genes **p1** and **p2** represent the absence of pigment. Their various combinations produce different shades of skin as shown in the following table:

No.	Shade of skin	Genotype
1	Full black	$P^1P^1P^1P^1$
2	Sambo	$P^1P^1P^2P^2$ or $P^1p^1P^1p^1$
3	Mulatto	$P^1P^1p^2p^2$ or $P^1p^1P^2P^2$ or $P^1p^1P^2p^2$
4	Quadroon	$P^1p^1p^2p^2$ or $p^1P^1P^2p^2$ or $P^1p^1P^2p^2$
5	White	$p^1p^1p^2p^2$

There are many traits in man which involve more than two pairs of cumulative genes. More the number of genes involved in a trait, the greater is the variety of its expression.

Pleiotropism

Pleiotropism is defined as the effect of a single gene **upon two or more characters which are not related.** In Drosophila the recessive gene for vestigial wings (**vg**) produces vestigial wings in homozygous condition. In addition to wing length it is also responsible for the production of (a) the tiny wing like balancer behind the wings; (b) certain bristles, (c) the structure of the spermatheca and (d) low number of eggs. This phenomenon of **multiple effect of a single gene is called pleiotropism.**

Lethal Genes

Genes may affect viability as well as visible traits of the organism. **A lethal gene is that which produces an effect which differs from the normal condition that its possessor is unable to survive.** A lethal gene kills its possessor. Thus the victims of lethal genes do not live to reproduce. Such genes are constantly propagated through heterozygous individuals.

Lethal genes in Man: Different lethal genes differ in time at which they bring about their lethal effects; the time depends upon the development and functioning of the structures, the abnormalities of which cause death. There is a mutant recessive gene in man which causes internal adhesions of the lungs. A child homozygous for the lethal gene might be able

to survive at embryonic development, but at birth, when it suddenly becomes dependent upon its lungs for its oxygen supply, it would die because its lungs could not expand properly. Being recessive, it could be carried by normal parents in the heterozygous condition without any ill effects.

The defects of the kidneys, lungs and digestive organs probably would not be lethal until birth, for these organs do not begin functioning until this time. So genes cause some types of heart defects would be lethal early in embryonic life.

The infants with **juvenile amaurotic idiocy** disease lose eyesight between the ages of 4 and 7. Mental and physical powers deteriorate and they die before adolescence. This disease is due to a recessive gene in homozygous condition. **Infantile amaurotic idiocy** is another dangerous disease in infants. It is also due to the presence of a particular recessive gene in homozygous condition. **Sickle cell anemia** is another disease caused by a recessive lethal gene in homozygous condition.

Lethal genes in mice: In the house mouse yellow coat colour is due to a gene which is dominant to all genes for other coat colours like black, white etc. It is further known that all yellow individuals are heterozygous (**Yy**). When two yellow individuals are bred together, they always produce offsprings in the ratio of 2 heterozygous yellow and 1 non-yellow. But the expected ratio is 1 pure yellow; 2 heterozygous yellow and 1 non-yellow. The actual ratio 2:1 is an unusual ratio and is due to the fact that the homozygous yellow individuals (**YY**) die in the embryonic stages. The gene dominant in mouse, is thus lethal, causing death in the homozygous condition.

Parents : Yellow **Yy** x Yellow **Yy**

Gamete : **Y** **y** **Y** **y**

Offspring: Homozygous Yellow **YY** (Dies) — Heterozygous yellow **Yy** — Homozygous **yy** non yellow

Fig. 27: Lethal genes in mice.

Sex linked lethals: A lethal gene present in a sex chromosome (X) is called a sex linked lethal gene. In **Drosophila**, sometimes 50% of the males in a small population are missing. So the normal sex ratio 1:1 is modified and becomes 2 females and 1 male. It is due to sex linked lethal gene. Some of the females may contain a recessive lethal gene in one of

Parents : Rose comb male **RRpp** x Pea comb female **rrPP**

Gametes : Rp, rP

F1 Generation : **RrPp** Walnut

F1 Fowls are crossed. Walnut male **RrPp** x Walnut female **RrPp**

Gametes : RP, Rp, rP, rp, RP, Rp, rP, rp

Gametes	RP	Rp	rP	rp
RP	**RRPP** Walnut	**RRPp** Walnut	**RrPP** Walnut	**RrPp** Walnut
Rp	**RRPp** Walnut	**RRpp** Rose	**RrPp** Walnut	**Rrpp** Rose
rP	**RrPP** Walnut	**RrPp** Walnut	**rrPP** Pea	**rrPp** Pea
rp	**RrPp** Walnut	**Rrpp** Rose	**rrPp** Pea	**rrpp** Single

F2 generation: Walnut : 9, Rose : 3, Pea : 3, Single : 1

Fig. 17: Inheritance of combs.

Epistasis

Epistasis is a Greek word which means **stopping** or **suppression.** In some animals a gene at one locus on a chromosome suppresses or masks the expression of a gene at another locus. Such genes are known as **inhibiting genes,** since the

If any one of the wild genes of this series becomes mutant then the nature of the end product is altered. In other words, the mutant genes cause **metabolic blocks** in synthetic pathway. If the genotype is homozygous for the recessive gene 3 (**cc**), then the biosynthetic pathway ends with metabolite **K**. The gene 4 has no power to convert the metabolite **K** into melanin. So the metabolite **K** is accumulated and it leads to abnormal phenotype (albino) without melanin pigment. Thus the recessive alleles of the gene 3 mask the phenotypic expression of the gene 4. The gene which suppresses or masks the expression of a gene at another locus is termed **epistatic** and the gene which is suppressed by an epistatic gene is called **hypostatic** gene.

Initial Substance
↓ ←Enzyme 1 ←—— Gene 1 **AA** or Aa
J-Metabolite
↓ ←Enzyme 2 ←—— Gene 2 BB or Bb
K-Metabolite
↓ ←Enzyme 3 is not synthesized, Gene 3 **CC** is **mutated into cc, Albino.**

Further conversions are blocked. Melanin pigments are not produced. Albinism is resulted.

Fig.22: Biosynthetic pathway in mutants.

Duplicate genes (15:1)

When a single character is controlled by two or more pairs of **alleles** independently, the genes are called **duplicate genes.**

George H. SHULL (1914) reported a case of duplicate gene in the common weeds, shepherd purse **Bursa bursa pastoris.** There are two varieties of seeds. Normal variety produces a **triangular** seed case and the other mutant variety produces **oval** seed case. When these two varieties are crossed, the resulting F1 plants produce triangular seeds (which is thus dominant). When the F1 hybrids are selfed, in F2 plants with triangular seeds and oval seeds are produced in the ratio 15:1 instead of the normal Mendelian ratio.

G.H.SHULL reported that two different independently segregating non-allelic duplicate dominant genes (**T** and **D**) influence the shape of the seed. In other words, two different non allelic genes **T** and **D** which are located at two different loci had the power to determine the triangular shape of the seed. Even a single gene **T** or **D** is enough to produce the the triangular shape. When both genes occur in the recessive condition, the oval shape is produced.

Shank of fowls: In poultry one variety of fowl, (the black leghorns) has feathered shanks and another variety (Buff Rocks) has featherless shanks. A cross between the two varieties produces the F1 with feathered shanks (dominant). In the F2 the fowls appear in the ratio 15:1 ie., 15 birds with feathered shanks and one with featherless shank. Thus this inheritance is also due to the presence of duplicate factors.

7. The colour of corn aleurone is known to be controlled by several genes; A, C and R are all necessary for colour to be produced. The locus of a dominant inhibitor of aleurone colour I is very closely linked to that of C. Thus any one or more of the genotypes I, aa, cc, or rr produces colourless aleurone.

(a) What would be the coloured : colourless ratio among F2 progeny from the Cross AAIICCRR x aaiiCCRR ? (b) What proportion of the colourless F2 is expected to be homozygous ?

4

Multiple Alleles

Mendel proposed that a character is controlled by a single pair of genes. The two genes of a character are located in the same locus of homologous chromosomes. These two genes are called **alleles.** Alleles are a pair of genes controlling the same character and located at the same locus of the homologous chromosomes. These alleles undergo mutation to give rise to three or more alleles located in the same locus of the homologous chromosomes. These mutant alleles express different alternatives of the same character. Such genes are called multiple alleles. Multiple alleles may be defined as a series of three or **more genes which control the same character and occupy the same locus in the homologous chromosomes.** The multiple alleles arise by the mutation of normal genes.

A set of multiple alleles may contain **three, four** or **more members.** All the members occupy the same locus in the homologous chromosomes since all these alleles are formed by the mutation of one gene. Out of several allelic forms of a gene, **a given diploid individual possesses any two alleles of the allelic series and its gamete carries only one allele.**

1. Coat colour in rabbits

In rabbits the coat colour is controlled by multiple alleles There are four varieties of rabbits. They are:

1. Agouti
2. Chinchilla
3. Himalayan and
4. Albino.

1. **Agouti:** This is the wild type rabbit and its body is **brownish grey** in colour. The dominant gene **C** is responsible for the brown coat colour. This dominant gene

32 Multiple Alleles

A set of three or more alleles of the same gene on the homologous chromosomes to control a particular trait is called ***multiple alleles.*** They occupy a particular locus on the homologous chromosomes. Each of these alleles is a repetition of the original sequence with slight changes. For example, gene for asynaptic meiosis in 8th chromosome of tomato exists in the form of five alleles (as *1* or as *2* or as *3* or as *4* or as *5*)

A set of multiple alleles may contain ***three, four*** or ***more members.*** All the alleles occupy the same locus in the homologous chromosomes, since all these alleles are formed by the mutation of one gene. Out of several allelic forms of a gene, ***a given diploid individual possesses any two alleles of the allelic series and its gamete carries only one allele.***

The ***ABO Blood group*** is controlled by multiple alleles.

The following are the examples for multiple alleles:

1. *ABO blood group*
2. *Rh blood group*
3. *Nature of wings in Drosophila*
4. *Coat colour in rabbit*
5. *Self sterility in tobacco.*

33 ABO Blood Group

Landsteiner found four types of human beings depending on the presence or absence of antigens in the blood. They are ***A, B, AB,*** and ***O***.

A group persons contain antigen ***A*** on the RBC.

B group persons contain antigen ***B*** on the RBC.

AB group persons contain both antigens ***A*** and ***B*** on the RBC.

O group persons contain ***no antigens*** on the RBC.

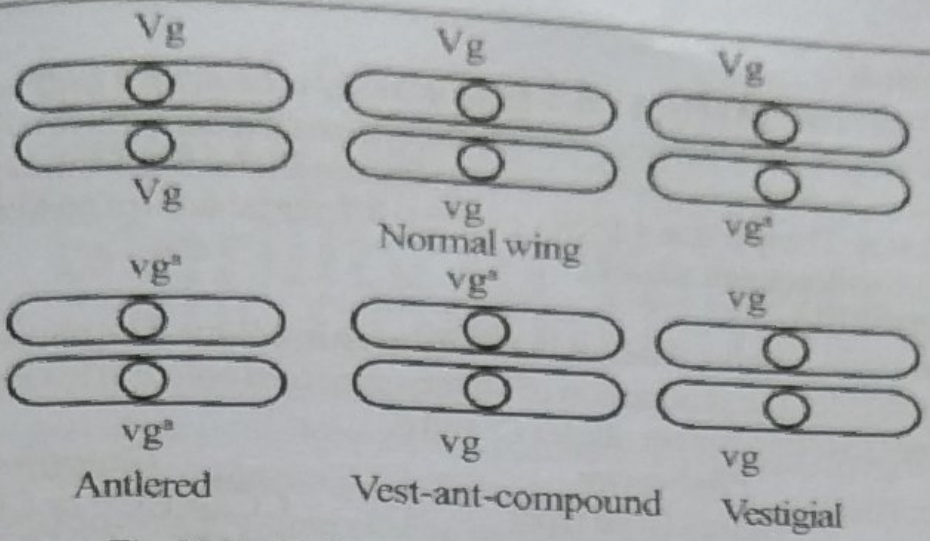

Fig.35.3: Multiple alleles of Drosophila.

Parents : Vestigial x Antlered
vg vg ↓ **vg^a vg^a**
F$_1$: **vgvg^a**
Vestigial antlered compound
F$_1$ Crossed : **vg vg^a** x **vg vg^a**
F$_2$ Genotypic ratio : **1vgvg : 2vg^a vg^a : 1 vg^a vg^a**
Phenotypic ratio: ***1 Vestigial: 2 Vest-ant-compound: 1 Antlered***

Fig:35.4: Cross between Drosophila with vestigial wing and antlered wing.

36 Multiple Alleles- Coat Colour in Rabbits

In rabbits, the coat colour is controlled by multiple alleles. There are four varieties of rabbits. They are:

1. *Agouti*
2. *Chinchilla*
3. *Himalayan* and
4. *Albino.*