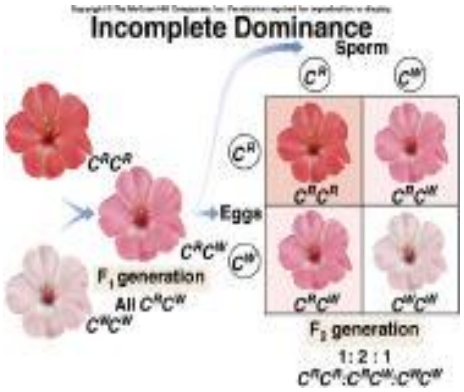
Variation in Dominance Relation

Some cases the phenotypes of heterozygotes are found to be different from either of the homozygotes :

1. Incomplete Dominance

Sometimes in a heterozygote dominant allele does not completely mask the phenotypic expression of the recessive allele and there occurs an intermediate phenotype in the heterozygote. This is called **incomplete dominance**.



Examples. 1. When a red flowered pea plant (RR) is crossed with white flowered pea plant (rr) then the F1 hybrid pea plants are found to have pink flowers. It shows that gene for red colour could not completely dominate the gene for **white** colour as shown in the figure

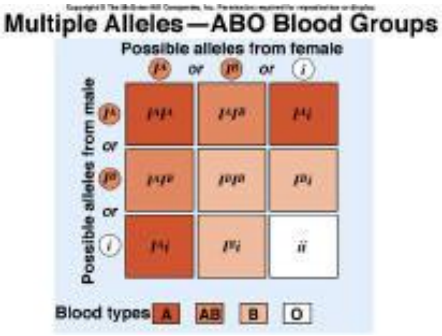
. In such a case, F2 phenotypic ratio and genotypic ratio are the same, as follows :

F2 phenotypic ratio = 1 Red : 2 Pink : 1 White

F2 genotypic ratio = 1 RR : 2 Rr : 1 rr

2. Codominance

Sometimes both alleles of a gene in a heterozygote lack the dominant and recessive relationship, *i.e.*, each allele is capable of some degree of phenotypic expression. In a sense, codominance is no dominance at all, the heterozygote showing the phenotypes of both homozygotes. Hence, heterozygote genotype gives rise to a phenotype distinctly different from either of the homozygous genotypes.



Ex. The alleles governing the **ABO blood group** system in humans are codominants and may Blood groups actually represent is a pattern of inheritance in which both alleles of a gene are fully expressed. A person with AB blood has both A and B antigens on their red blood cells. With codominance, both alleles produce an effective product.

Deviation from mendel's dihybrid phenotypic ratio

The Mendelian dihybrid phenotypic ratio of 9 : 3 : 3 : 1 is obtained only when the alleles at both gene loci display dominant and recessive relationship. If one or both gene loci have incompletely dominant alleles, or codominant alleles or lethal alleles, the dihybrid ratio becomes modified variously, such as follows :

Ex 1.- 3 : 6 : 3 : 1 : 2 : 1 Ratio

Ex 2.- 1 : 2 : 1 : 2 : 4 : 2 : 1 : 2 : 1 Ratio

Genetic Interaction (Epistasis)

A gene or locus which suppressed or masked the action of a gene at another locus was termed **epistatic gene.**

Difference Between Dominance and Epistasis

The phenomenon of dominance involves **intra-allelic** gene suppression, or **the masking effect which one allele has upon the expression of another allele at the same locus**, while the phenomenon of epistasis involves **inter-allelic** gene suppression or the masking effect which **one gene locus has upon the expression of another**. The classical phenotypic ratio of 9 : 3 : 3 : 1 observed in the progeny of dihybrid parents becomes modified by epistasis into ratios which are various combinations of the 9: 3 : 3 : 1 groupings.

Kinds of epistatic interaction

When in dihybrid crosses, the epistatic interactions occur between two genes, less than four phenotypes appear in F2. Such bigenic (two gene) epistatic interactions may be of following six types: such as

1. **Dominant Epistasis (12 : 3 : 1)**
2. **Recessive Epistasis (9 : 3 : 4)**

Lethal genes are mutant genes and result in the death of the individual which carries them. Death of the individual occurs either in the prenatal or postnatal period prior to sexual maturity. A **fully** (completely) **dominant lethal allele** kills both in homozygous and heterozygous states. Individuals with a dominant lethal allele die before they can leave progeny. Therefore, the mutant dominant lethal is removed from the population in the same generation in which it arose. **Recessive lethal genes** kill only when they are in a homozygous state and they may be of two kinds :

1. one which has no obvious phenotypic effect in heterozygotes .
2. one which exhibits a distinctive phenotype when heterozygous.

Lethal alleles in human beings. In humans several hereditary diseases have lethal effects. Few important lethal genes of man are following :

1. **Congenital ichthyosis.** One of the most typical cases of a recessive lethal gene in man is expressed in congenital ichtyosis. At birth children afflicted with this disease have a crusted leathery skin with deep fissures down to the subcutaneous tissue; the fissures lead to bleeding, infection and death. Congenital ichthyosis occurs only when there occurs homozygous condition for its recessive lethal genes.

2. **Infantile amaurotic idiocy.** A recessive allele in homozygous condition causes a fatal disease called **infantile amaurotic idiocy** in juvenile stage. Bearers of this genotype begin to lose their eye sight between the age of four to seven years. The complete blindness is followed by mental degeneration and finally death before adolescence.

3. **Thalassemia** or **Cooley's anaemia** is a haemoglobin disease somewhat similar to sickle cell anaemia. It occurs mostly in children (in India and other countries such as Italy, Greece and Syria) and is nearly 100 per cent fatal (lethal). Thalassemia is

controlled by a single gene c which in homozygous condition (cc), produces the severe Cooley's anaemia or **thalassemia major** and causes death of the patient. The heterozygous condition of this lethal gene (Cc) results in a mild form of the disease called **thalasemia minor** or **microcythemia**

Penetrance

The ability of a given gene or gene combination to be expressed phenotypically to any degree is called **penetrance**. It is of following two kinds :

1. Complete Penetrance

Most dominant and recessive genes in homozygous conditions and many completely dominant genes even in heterozygous conditions give their complete phenotypic expressions. Such genes are called to have **complete penetrance**.

2. Incomplete Penetrance

Some genes in homozygous as well as in heterozygous conditions fail to provide complete (cent per cent) phenotypic expression of them. Such genes are called to have **incomplete penetrance**.

Examples of Incomplete Penetrance

(i) **Polydactyly** in man is thought to be produced by a dominant gene P. The normal condition with five digits on each limb is produced by the recessive genotype (pp). Some heterozygous individuals (Pp) are not polydactylus and, therefore, has a penetrance of less than 70%.

(ii) In man, the tendancy to develop **diabetes mellitus** (a condition in which there is an excess of sugar in the blood) is controlled by certain genes. However, not everyone carrying the genes for diabetes actually develops the condition, for these genes have incomplete penetrance.

Effects of environment on penetrance.

The environmental factors and genetical background have some definite effect on the degree of penetrance of a gene. For example, when various twins which carry genes for diabetes mellitus are studied, it is found that the disease appears only in those cases which ate more carbohydrate foods (starch and sugars).

Expressivity

A trait though penetrant, may be quite variable in its phenotypic expressions. The degree of effect produced by a penetrant genotype is called **expressivity**.

Example of expressivity. In man the polydactylous condition may be penetrant in the left hand (6 fingers) and not in the right (5 fingers); or it may be penetrant in the feet and not in the hands.

Effects of environment on expressivity.

The expressivity of a given gene is often influenced by environmental conditions. Examples of environmental effects on the expressivity of a gene includes such cases as the differences in the severity of symptoms of an inheritable allergy, or the differences in height

of identical twins who have been raised in different home (with different diets), or who have had different medical histories (one with a serious childhood disease, the other escaping this disease).

Pleiotropism (pleiotropy)

Up till now we have observed that a specific gene has a specific effect upon a specific phenotypic trait or in other words, each gene (allele) has its relation with a single phenotypic trait, but, this is not the case. A single gene often influences more than one phenotypic trait. However, it may be that one gene may cause evidently well marked expression of some phenotypic trait (**major effect**) then the others with less evident phenotype (**secondary effect**). Most genes have their multiple effects and are called **pleiotropic genes**. The phenomenon of multiple effect (multiple phenotypic expressions) of a single gene is called **pleiotropism**.

Example of Pleiotropism

** In human, the gene for disease **phenylketonuria** has pleiotropic effect and produces various abnormal phenotypic traits, collectively called **syndrome**. For example, the affected individuals secrete excessive quantity of amino acid phenylalanine in their urine, cerebrospinal fluid and blood. They become short stature, mentally deficient, with widely spaced incisors, with pigmented patches on skin, with excessive sweating, and with non-pigmented hairs and eyes.

Quantitative Genetics

(Inheritance of Multiple Genes)

The phenotypic traits of the different organisms may be of two kinds, qualitative and quantitative. The **quali tative traits** are the classical Mendelian traits of **kinds** such as form antigens and antibodies (*e.g.*, blood group types of man) The organisms possessing qualitative traits have distinct (separate) phenotypic classes and are said to exhibit **discontinuous variations**.

The **quantitative traits**, however, are economically important measurable phenotypic traits of **degree** such as height, weight, shape, skin pigmentation, metabolic activity, reproductive rate, behaviour, eye-facet, susceptibility to pathological diseases or intelligence in man; amount of flowers, fruits, seeds, milk, meat or egg. They do not show clear cut differences between individuals and forms a spectrum of phenotypes which blend imperceptively from one type to another to cause **continuous variations**.

The inheritance of polygenes or quantitative traits is called **quantitative inheritance**, **multiple factor inheritance**, **multiple gene inheritance** or **polygenic inheritance**.

Characteristics of multiple genes

Multiple genes for quantitative traits have following characteristics :

1. Each contributing allele in the series of multiple genes produces an equal effect.
2. Effects of each contributing allele are cumulative or additive.
3. There is no dominance, rather, there exist pairs of contributing and non-contributing alleles.
4. There is no epistasis (masking of the phenotypes) among genes at different loci.
5. There is no linkage involved.

6. The environmental conditions have considerable effect on the phenotypic expression of poly- genes for the quantitative traits.

Skin Colour in Man:

The classical example of polygenic inheritance was given by **Davenport** (1913). He found that two pairs of genes, A-a and B-b cause the difference in skin Pigmentation between negro and Caucasian people. These genes were found to affect the character in additive fashion. Thus, a true negro has four dominant genes, AABB, and a white has four recessive genes aabb. The F1 offspring of mating of aabb with AABB, are all AaBb and have an intermediate skin colour termed **mulatto**. A mating of two such mulattoes produces a wide variety of skin colour in the offspring, ranging from skins as dark as the original negro parent to as white as the original white parent. The result of this cross have been shown in Figure .The F2 phenotypicand genotypic ratios have been tabulated in Table .

P1 : AABB × aabb
Negro White

P1 gametes : AB ab

F1 : AaBb
Mulattoes
Intermediate skin colour

Intercross : Aa Bb × AaBb
Mulattoes Mulattoes

& \ %	AB	Ab	aB	ab
AB	AABB Like negro	AABb Darker than mulattoes	AaBB Darker than mulattoes	AaBb Like mulattoes
Ab	AABb Darker than mulattoes	AAbb Like mulattoes	AaBb Like mulattoes	Aabb Lighter than mulattoes
aB	AaBb Darker than mulattoes	AaBb Like mulattoes	aaBB Like mulattoes	aaBb Lighter than mulattoes
ab	AaBb Like mulattoes	Aabb Lighter than mulattoes	aaBb Lighter than mulattoes	aabb Like white

F_2 :	Negro colour (1/16)	Colour between mulattoes and negro (4/16)	Colour of mulattoes (6/16)	Colour between mulattoes and white (4/16)	White skin colour (1/16)

5.3. Checkerbord exhibiting segregation of skin colour in F_2 generation from a marriage between a negro and a white person.

2. Eye Colour in Man

In human beings, the colour of eye is found to be determined by polygenes. These genes have been suggested to be X-linked . At least 9 classes of eye colour can be recognized in humans. In order of increasing amount of melanin pigmentation, these eye colours can be designated as light blue, medium blue, dark blue, grey, green, hazel, light brown, medium

brown and dark brown. The number of contributing alleles for these colours have been tabulated in Table 5-4.

Table 5.4. **Number of contributing alleles for each type of eye colour of human beings (Source : Burns and Bottino, 1989).**

Number of contributing alleles	Eye colour
0	Light blue
1	Medium blue
2	Dark blue
3	Grey
4	Green
5	Hazel
6	Light brown
7	Medium brown
8	Dark brown