

M.Sc. Microbiology, 2nd Semester MCB 202 : Genetics and Gene regulation Gr. A: Fundamental Genetics

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Introduction

The phenomenon of two or more genes affecting the expression of each in various ways in the development of a single character of an organism is known as gene interaction.

Most of the character of living organisms are controlled/influenced/governed by a collaboration of several different genes.

Mendel assumed that characters are governed by single genes but later it was discovered that many characters are governed by two or more genes.

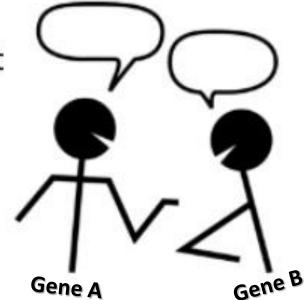
GENE INTERACTION

Types of gene interaction-

Gene-Gene and Gene-Environment

Interaction between allelic genes

- Complete dominance
- Incomplete dominance
- Co-dominance

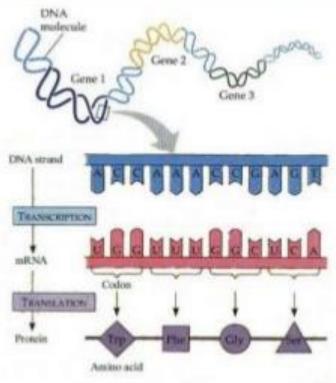


Interaction between non-allelic genes

- Additive gene interaction
- Duplicate gene
- Dominant suppression
- Dominant epistasis
- Recessive epistasis

MECHANISM OF GENE

- Genes are segments of DNA that code for particular polypeptide in the form of a specific sequence of its base pair.
- Proteins are the end products
 of gene expression, and so gene
 interactions are interactions between proteins that are controlled by these genes.



Interaction between Allelic genes

Complete Dominance



- When two unlike alleles responsible for a single character are present in a single individual, one allele can mask the expression of another allele. That is, one allele is dominant to the other. The latter is said to be recessive.
- In complete dominance, the effect of one allele in a heterozygous genotype completely masks the effect of the other. The phenotype of the heterozygote is indistinguishable from that of the dominant homozygote.

Each true-breeding plant of the parental generation has identical alleles, *PP* or *pp*.

one allele for the flower-color gene. In this case, every gamete produced by one parent has the same allele.

Gametes (circles) each contain only

Union of the parental gametes produces F₁ hybrids having a Pp combination. Because the purpleflower allele is dominant, all these hybrids have purple flowers.

When the hybrid plants produce gametes, the two alleles segregate, half the gametes receiving the P allele and the other half the p allele.

This box, a Punnett square, shows all possible combinations of alleles in offspring that result from an $F_1 \times F_1$ (*Pp* × *Pp*) cross. Each square represents an equally probable product of fertilization. For example, the bottom left box shows the genetic combination resulting from a *p* egg fertilized by a P sperm.

Random combination of the gametes results in the 3:1 ratio that Mendel observed in the F_2 generation.

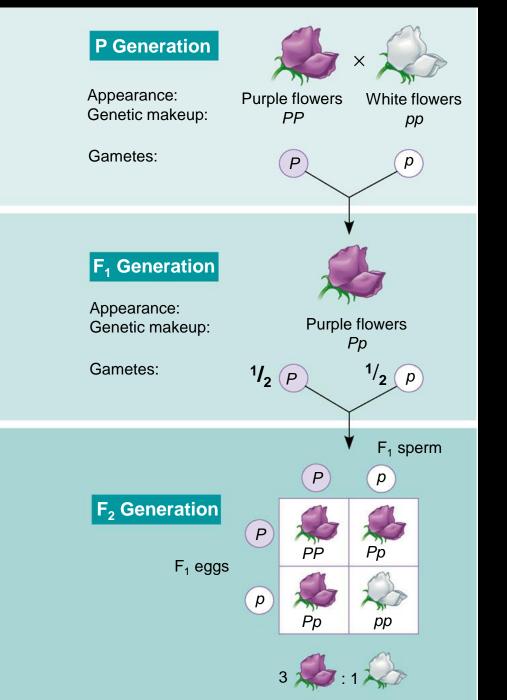


Table 14.1 The Results of Mendel's F1 Crosses for SevenCharacters in Pea Plants					
Character	Dominant Trait	×	Recessive Trait	F ₂ Generation Dominant:Recessive	Ratio
Flower color	Purple	×	White	705:224	3.15:1
Flower position	Axial	×	Terminal	651:207	3.14:1
Seed color	Yellow	×	Green	6022:2001	3.01:1
Seed shape	Round	×	Wrinkled	5474:1850	2.96:1
Pod shape	Inflated	×	Constricted	882:299	2.95:1
Pod color	Green	×	Yellow	428:152	2.82:1
Stem length	Tall	×	Dwarf	787:277	2.84:1

Incomplete dominance

- One exception to Mendel's rules is that one allele is always completely dominant over a recessive allele. Sometimes an individual has an intermediate phenotype between the two parents, as there is no true dominant allele. This pattern of inheritance is called **incomplete dominance**.
- Incomplete dominance occurs when the phenotype of the heterozygous genotype is distinct from and often intermediate to the phenotypes of the homozygous genotypes.
- For example, the snapdragon flower color is either homozygous for red or white. When the red homozygous flower is paired with the white homozygous flower, the result yields a pink snapdragon flower. The pink snapdragon is the result of incomplete dominance.

 When plants of the F1 generation are selfpollinated, the phenotypic and genotypic ratio of the F2 generation will be 1:2:1 (Red:Pink:White) for both generations



Phenotype	Red	Pink	White
Genotype	RR	Rr	rr

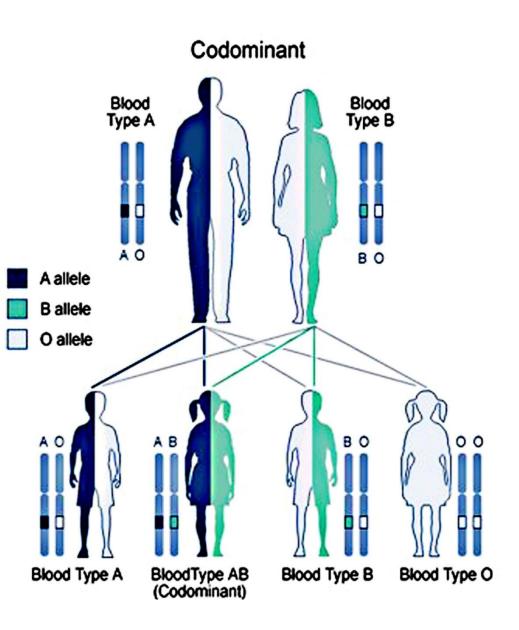
Co-dominance

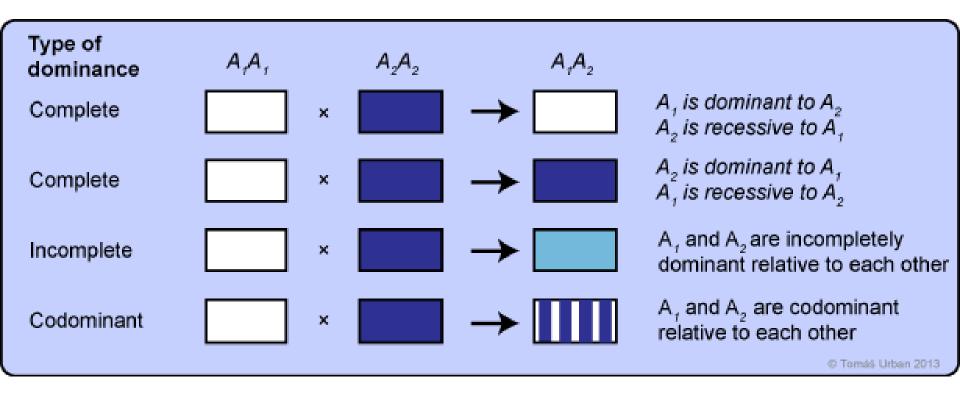
- Co-dominance occurs when the contributions of both alleles are visible in the phenotype.
- A condition in which the alleles of a gene pair in a heterozygote are fully expressed.
- Resulting in offspring with a phenotype that is neither dominant nor recessive, rather combination of two.

ABO blood group: Co-dominance & Multiple Alleles

- ABO blood group system, chemical modifications on the surfaces of blood cells are controlled by <u>three alleles (I^A, I^B and I^O) at the ABO locus.</u>
- Two alleles are dominant and completely expressed (designated I^A and I^B), while one allele is recessive (I^O).
- The I^A allele encodes for red blood cells with the A antigen, while the I^B allele encodes for red blood cells with the B antigen.
- The I^A and I^B alleles produce different modifications, and the non-functional I^O allele produces no modification.

I^A and I^B alleles are each dominant to I^O. I^AI^A and I^AI^O individuals both have type A blood, and I^B I^B and I^BI^O individuals both have type B blood. But I^AI^B individuals have both modifications on their blood cells and thus have type AB blood, so the I^A and I^B alleles are said to be co-dominant.





Interaction between Non-allelic genes





(a)





(c)

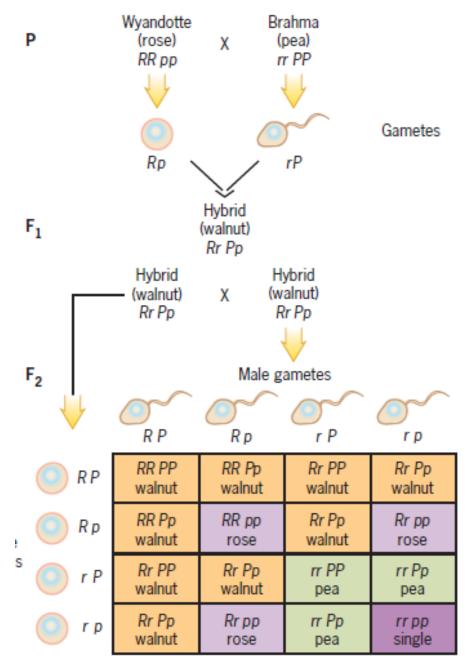
(d)

GENE INTERACTIONS

Some of the first evidence that a trait can be influenced by more than one gene was obtained by Bateson and Punnett from breeding experiments with chickens. Their work was carried out shortly after the rediscovery of Mendel's paper. Domestic breeds of chickens have different comb shapes : Wyandottes have "rose" combs, Brahmas have "pea" combs, and Leghorns have "single" combs. Crosses between Wyandottes and Brahmas produce chickens that have yet another type of comb, called "walnut." Bateson and Punnett discovered that comb type is determined by two independently assorting genes, R and P, each with two alleles Wyandottes (with rose combs) have the genotype RR pp, and Brahmas (with pea combs) have the genotype rr PP. The F₁ hybrids between these two varieties are therefore Rr Pp, and phenotypically they have walnut combs. If these hybrids are intercrossed with each other, all four types of combs appear in the progeny: 9/16 walnut (R-P-), 3/16 rose (R-pp), 3/16 pea (rr P-), and 1/16 single (rr pp). The Leghorn breed, which has the single-comb type, must therefore be homozygous for both of the recessive alleles.

The work of Bateson and Punnett demonstrated that two independently assorting genes can affect a trait. Different combinations of alleles from the two genes resulted in different phenotypes, presumably because of interactions between their products at the biochemical or cellular level.

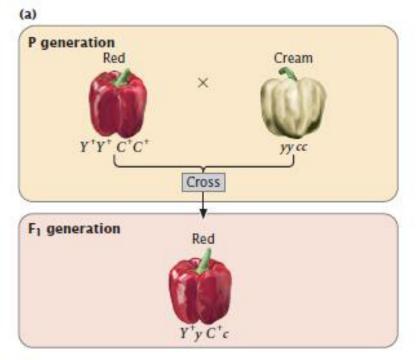
Comb shapes in chickens of different breeds. (a) Rose, Wyandottes; (b) pea, Brahmas; (c) walnut, hybrid from cross between chickens with rose and pea combs; (d) single, Leghorns.



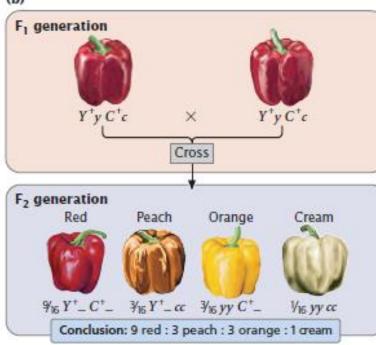
Summary: 9/16 walnut, 3/16 rose, 3/16 pea, 1/16 single

i.

c



(b)



Genotype	Phenotype
$Y^{+}_{C^{+}}_{C^{+}}}}}}}}}}}}}}}}}}}}}}}}}}}}}}}}}}}$	red
<i>Y</i> ⁺ _ <i>cc</i>	peach
уу C ⁺ _	orange
уу сс	cream

Color in peppers of *Capsicum annuum* results from the relative amounts of red and yellow carotenoids, compounds that are synthesized in a complex biochemical pathway. The *Y* locus encodes one enzyme (the first step in the pathway), and the *C* locus encodes a different enzyme (the last step in the pathway). When different loci influence different steps in a common biochemical pathway, gene interaction often arises because the product of one enzyme affects the substrate of another enzyme.

Complementary Gene Interaction

- Type of interaction in which both dominant pair of genes have same phenotypic effect when present alone.
- But when present together produces different effect.
- Observed in flower colour of garden pea.
- Two varieties having white flower when crossed; F1 - Purple colour.
- Flower colour in pea is governed by two different pair of genes.
- 'C' produce white flower in one variety
- 'W' produce white flower in other variety
- C & W present together produces Purple

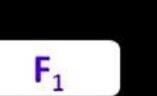


White Flower



ccWW

White Flower



CcWw

Purple Flower





-		
	3	
-	/	

	CW	Cw	cW	cw
CW	CCWW	CCWw	CcWW	CcWw
Cw	CCWw	CCww	CcWw	Ccww
cW	CcWW	CcWw	ccWW	ccWw
cw	CcWw	Ccww	ccWw	ccww

9 Purple : 7 White

Recessive gene factor act as complementary to each other, the interaction is known as complementary





Sweet pea (Lathyrus odoratus)

Duplicate gene interaction

- Two pair of non-allelic gene affect the same character in such a way that either one of them or both in dominant condition produces same phenotypic effect.
- Absence of both dominant factor result in the production of different phenotype.
- Observed in Awned & awnless character in rice.
- Awned condition dominant over awnless.

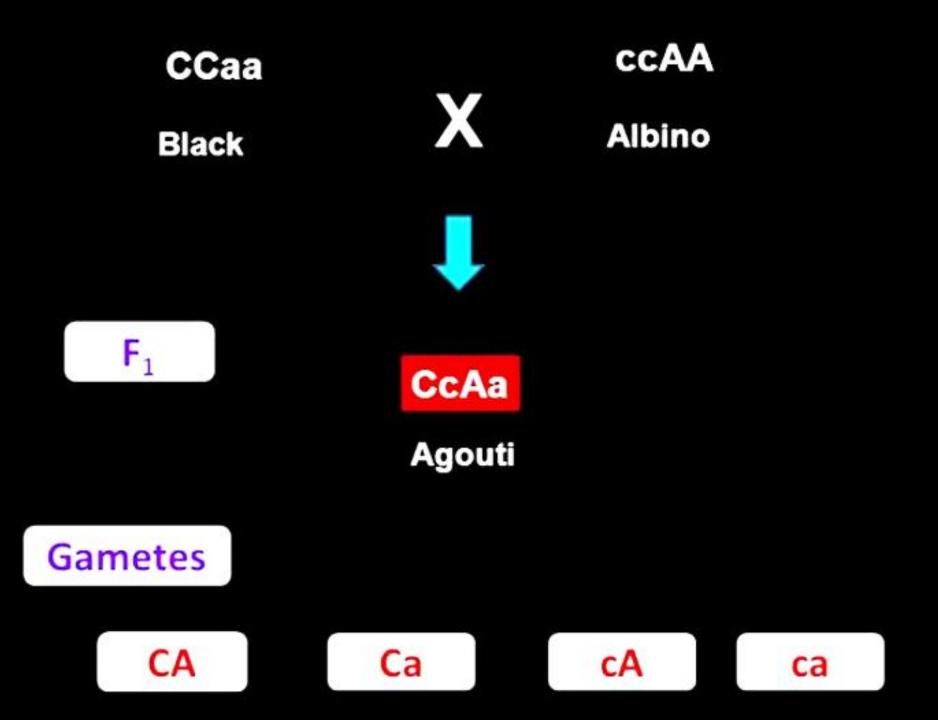




Supplementary Gene Interaction

- Type of interaction in which two pair of allele affect the same character.
- Out of two pair of genes; one of the dominant has visible effect itself; while other dominant gene express its effect when it is supplemented with other dominant gene.
- Noticed in coat colour in mice.
- 3 coat colour Black, Albino, Agouti
- Black dominant to Albino & Agouti







	CA	Са	cA	са
CA	CCAA	CCAa	CcAA	CcAa
Ca	CCAa	CCaa	CcAa	Ccaa
сA	CcAA	CcAa	ccAa	ccAa
са	CcAa	Ccaa	ccAa	ccaa

9 Agouti : 3 Black : 4 White

Gene 'C' necessary for development of Black Colour 'A' present in Albino mice 'A' is responsible for production of 'Agouti' in presence of 'C' 'A' alone has no effect- albino

Gene Interaction with Epistasis

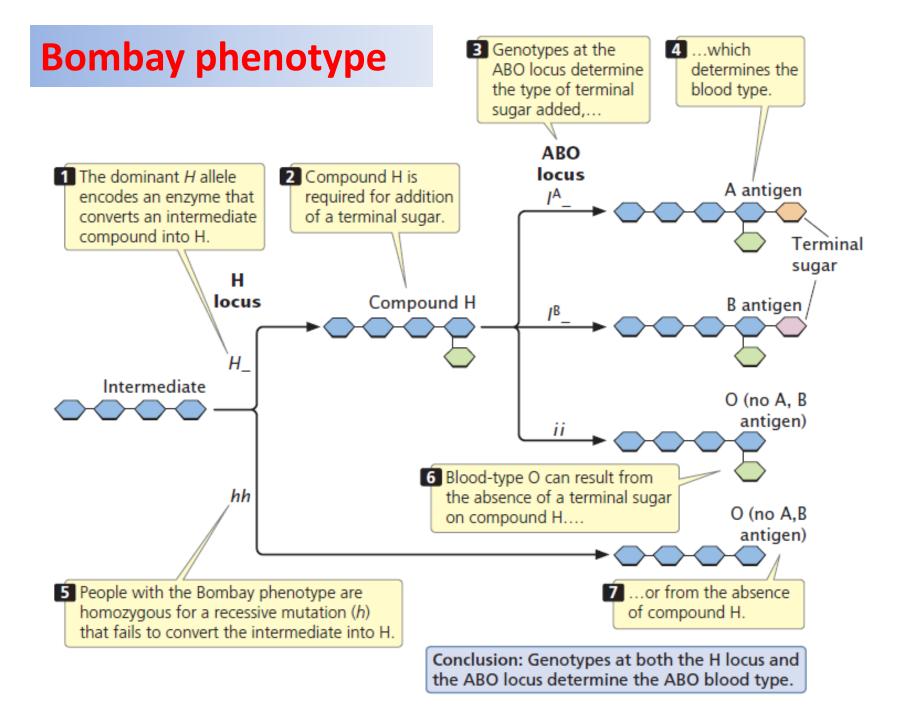
Sometimes the effect of gene interaction is that one gene masks (hides) the effect of another gene at a different locus, a phenomenon known as epistasis. In the examples of genic interaction that we have examined, alleles at different loci interact to determine a single phenotype. In those examples, one allele did not *mask* the effect of an allele at another locus, meaning that there was no epistasis. Epistasis is similar to dominance, except that dominance entails the masking of genes at the same locus (allelic genes). In epistasis, the gene that does the masking is called an epistatic gene; the gene whose effect is masked is a hypostatic gene. Epistatic genes may be recessive or dominant in their effects.

Recessive epistasis Recessive epistasis is seen in the genes that determine coat color in Labrador retrievers. These dogs may be black, brown, or yellow; their different coat colors are determined by interactions between genes at two loci

One locus determines the type of pig-

ment produced by the skin cells: a dominant allele B encodes black pigment, whereas a recessive allele b encodes brown pigment. Alleles at a second locus affect the *deposition* of the pigment in the shaft of the hair; dominant allele E allows dark pigment (black or brown) to be deposited, whereas recessive allele *e* prevents the deposition of dark pigment, causing the hair to be yellow. The presence of genotype ee at the second locus therefore masks the expression of the black and brown alleles at the first locus. The genotypes that determine coat color and their phenotypes are:

Genotype	Phenoty	ype	
<i>B</i> _ <i>E</i> _	black	If we cross a black Labrador homozygous for the domi-	
bb E_	brown	nant alleles (<i>BB EE</i>) with a yellow Labrador homozygous for the recessive alleles (<i>bb ee</i>) and then intercross the F_1 , we obtain progeny in the F_2 in a 9 : 3 : 4 ratio:	
B_ ee	yellow		
bb ee	yellow	obtain progeny in the 12 in a 2.5. Tradition	
		$\begin{array}{cccc} P & BB \ EE & \times & bb \ ee \\ & Black & Yellow \end{array}$	
		F_1 Bb Ee Notice that yellow dogs can carry alleles for either black or brown pigment, but these alleles are not expressed in their coat color.	
		Black Intercross	
		$F_2 = \frac{9}{16} B_E E_black$	
		$\frac{3}{16}$ bb E_ brown	
		$\left. \begin{array}{c} \frac{3}{16} B_ ee \text{ yellow} \\ \frac{1}{16} bb \ ee \text{ yellow} \end{array} \right\} \frac{4}{16} \text{ yellow}$	
copies o		a recessive epistatic allele, because two present to mask the expression of the nents. $B/_; E/_ b/b; E/_ ?/?; e/e$	

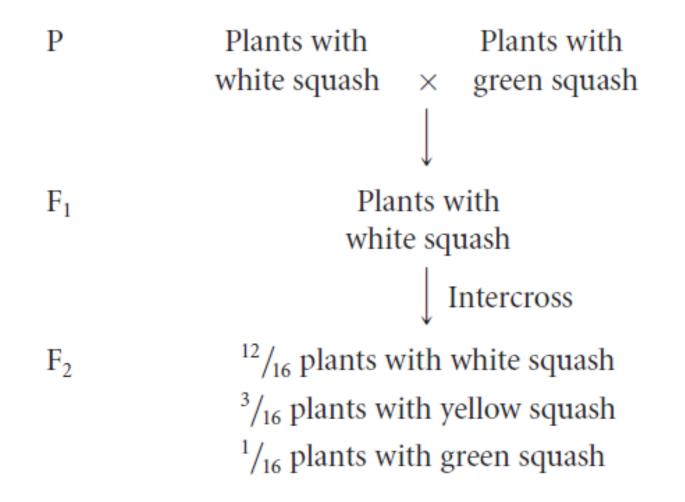


Genotype	H present	ABO phenotype
$H_I^A I^A, H_I^A i$	Yes	Α
$H_I^B I^B, H_I^B i$	Yes	В
$H_I^A I^B$	Yes	AB
H_ ii	Yes	0
hh I ^A I ^A , hh I ^A i, hh I ^B I ^B , hh I ^B i, hh I ^A I ^B , and hh ii	No	Ο

In this example, the alleles at the ABO locus are hypostatic to the recessive *h* allele.

Dominant epistasis In *recessive* epistasis, which we just considered, the presence of two recessive alleles (the homozygous genotype) inhibits the expression of an allele at a different locus. In *dominant* epistasis, only a single copy of an allele is required to inhibit the expression of the allele at a different locus.

Dominant epistasis is seen in the interaction of two loci that determine fruit color in summer squash, which is commonly found in one of three colors: yellow, white, or green. When a homozygous plant that produces white squash is crossed with a homozygous plant that produces green squash and the F_1 plants are crossed with each other, the following results are obtained:



How can gene interaction explain these results?

In the F_{2} , $\frac{12}{16}$, or $\frac{3}{4}$, of the plants produce white squash and $\frac{3}{16} + \frac{1}{16} = \frac{4}{16} = \frac{1}{4}$ of the plants produce squash having color. This outcome is the familiar 3 : 1 ratio produced by a cross between two heterozygotes, which suggests that a dominant allele at one locus inhibits the production of pigment, resulting in white progeny. If we use the symbol W to represent the dominant allele that inhibits pigment production, then genotype W_ inhibits pigment production and produces white squash, whereas ww allows pigment and results in colored squash.

Among those $ww F_2$ plants with pigmented fruit, we observe ${}^3/_{16}$ yellow and ${}^1/_{16}$ green (a 3 : 1 ratio). In this outcome, a second locus determines the type of pigment produced in the squash, with yellow (Y_{-}) dominant over green (yy). This locus is expressed only in ww plants, which lack the dominant inhibitory allele W. We can assign the genotype $ww Y_{-}$ to plants that produce yellow squash and the genotype ww yy to plants that produce green squash. The genotypes and their associated phenotypes are:

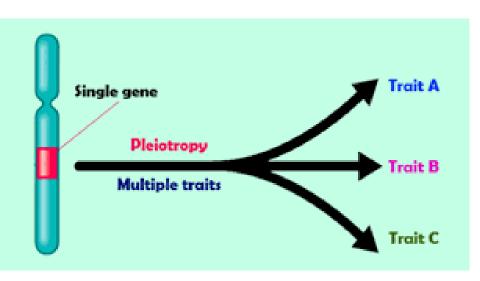
$W_Y_$	white squash
W_ yy	white squash
ww Y_	yellow squash
<i>ww yy</i>	green squash

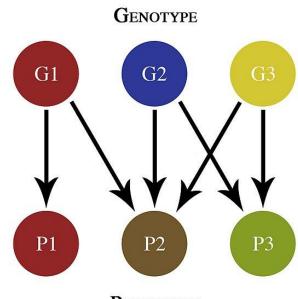


Pleiotropy

Pleiotropy occurs when one gene influences two or more seemingly unrelated phenotypic traits.

Therefore, a mutation in a pleiotropic gene may have an effect on several traits simultaneously due to the gene coding for a product used by a myriad of cells or different targets that have the same signaling function.





PHENOTYPE

Pleiotropy describes the genetic effect of a single gene on multiple phenotypic traits. The underlying mechanism is genes that code for a product that is either used by various cells or has a cascade-like signaling function that affects various targets.

Example:

- Phenylalanine is an amino acid that can be obtained from food. Phenylketonuria (PKU) causes this amino acid to increase in amount in the body, which can be very dangerous.
- The human disease is caused by a defect in a single gene on chromosome 12 that affects multiple systems.
- The most dangerous form of this is called classic PKU, which is common in infants. The baby seems normal at first but actually incurs permanent intellectual disability. This can cause symptoms such as mental retardation, abnormal gait and posture, and delayed growth. Because tyrosine is used by the body to make melanin (a component of the pigment found in the hair and skin), failure to convert normal levels of phenylalanine to tyrosine can lead to fair hair and skin.
- Phenylketonuria, results from a recessive allele; persons homozygous for this allele, if untreated, exhibit <u>mental</u> retardation, blue eyes, light skin color etc.

- Albinism is the mutation of the tyrosinase gene. This mutation causes the most common form of albinism.
- The mutation alters the production of melanin, thereby affecting melanin-related and other dependent traits throughout the organism.
- Melanin is a substance made by the body that is used to absorb light and provides coloration to the skin. Indications of albinism are the <u>absence</u> of color in an organism's eyes, hair, and skin, due to the lack of melanin.