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Genetics & Inheritance – Genetic Basis for Variation II

1. Introduction

Mendel's principles of inheritance only explain the most basic phenomena of inheritance. Other than Mendelian inheritance, you will also learn of non-Mendelian inheritance (e.g. autosomal linkage, epistasis), as well as how the environment plays a role in determining the phenotype of an organism. Statistical tests, such as the chi-squared test, which allow us to test the significance of differences between observed an expected results of genetic crosses will also be covered.

2. Learning Outcomes

x. use genetic diagrams to solve problems in dihybrid crosses, including those involving codominance, incomplete dominance, multiple alleles, sex linkage, autosomal linkage and epistasis

y. use genetic diagrams to solve problems including test crosses

z. explain the meaning of the terms linkage and crossing-over and explain the effect of linkage and crossing-over on the phenotypic ratios from dihybrid crosses

aa. describe the interaction between loci (epistasis) and predict phenotypic ratios in problems involving epistasis (knowledge of the expected ratio for various types of epistasis is not required; focus of this section is on problem solving)

cc. explain the difference between genetic variation that is continuous (many, additive genes control a characteristic) and genetic variation that is discontinuous (one or a few genes control a characteristic)

dd. use the chi-squared test to test the significance of differences between observed and expected results.

3. References

Campbell, N.A. and Reece, J.B. (2008). Biology, 9th edition. Pearson.

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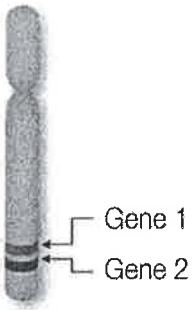
Contents

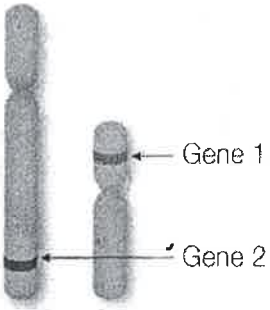
4. Autosomal Linkage

Notes to self

Linked genes refer to genes located on the **same chromosome / autosome**.



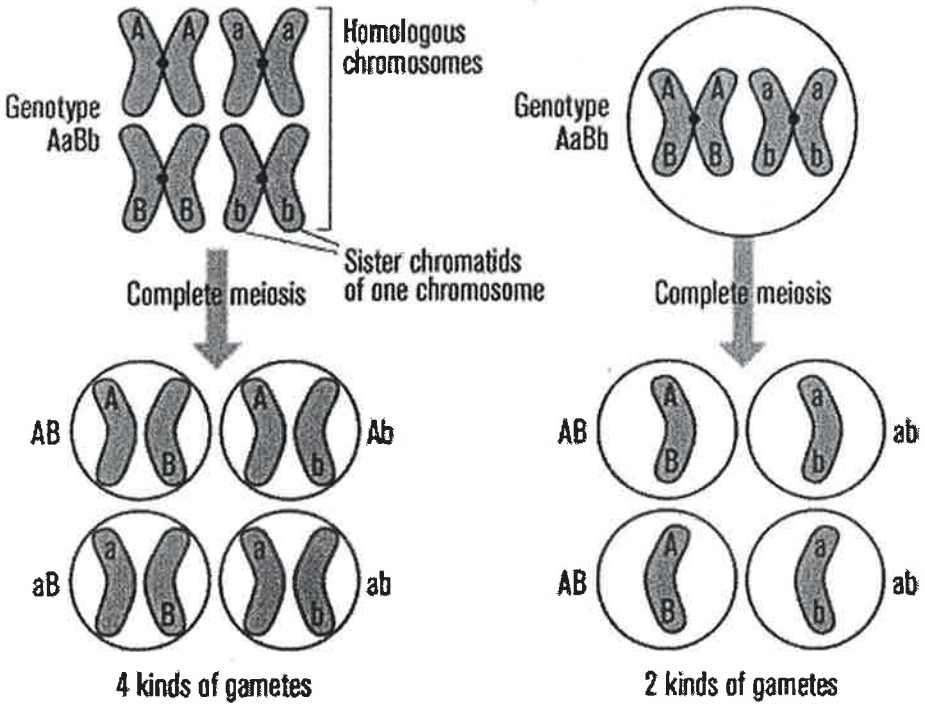
Genes 1 and 2 are <u>linked</u>



Genes 1 and 2 are <u>not linked</u>

As opposed to non-linked genes, linked genes are located close enough together on a chromosome that the alleles of the two genes **<u>tend</u> to be inherited together.**

Linked genes therefore **do not undergo independent assortment**.
(Note: chromosomes still undergo independent assortment, but not the genes.)



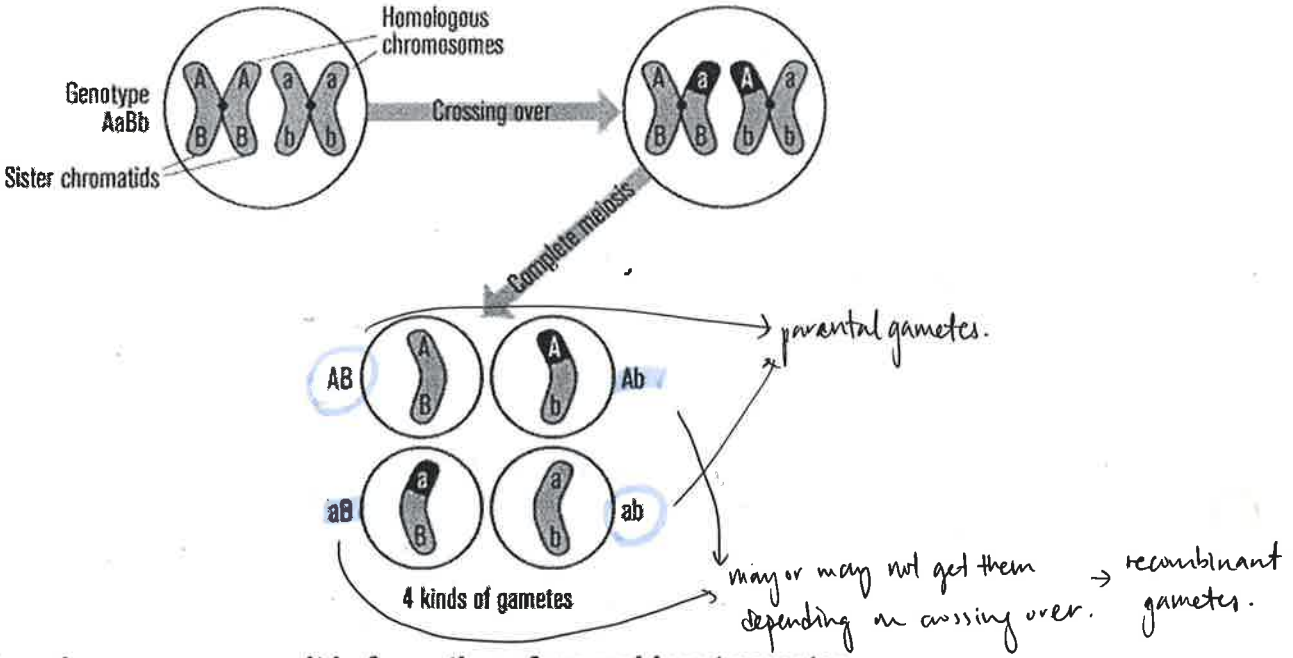
alleles A and B tend to be inherited tgt

alleles a & b tend to be inherited tgt.

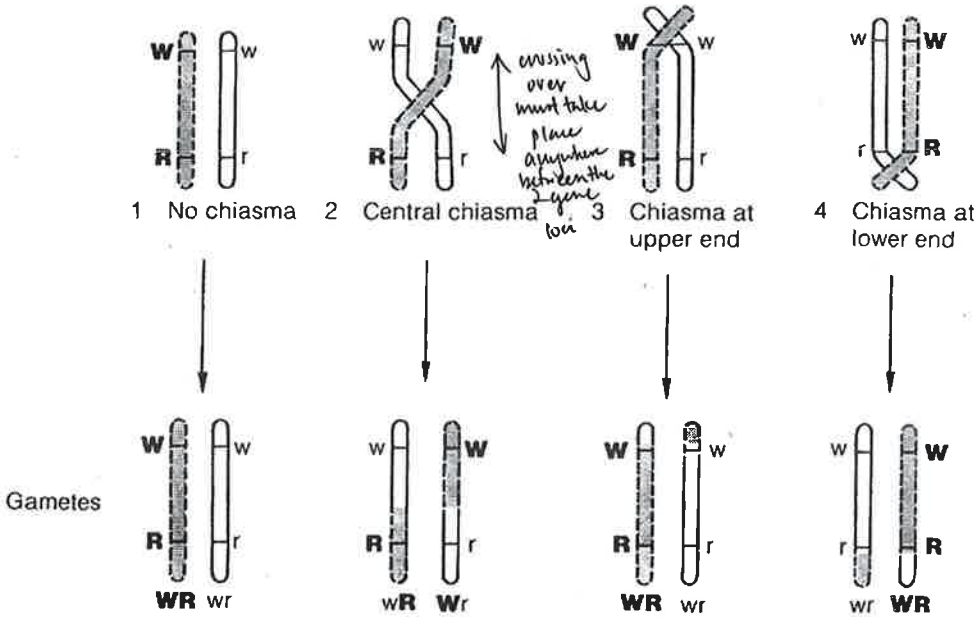
<u>Non-linked genes vs linked genes:</u>
<u>alleles of non-linked genes undergo independent assortment whereas alleles of linked genes tend to be inherited together and do not undergo independent assortment.</u>

However, the two alleles from the two genes on the same chromosome are not always inherited together. This happens when **crossing over** has taken place and **recombinants** are formed.

Notes to self



Crossing over can result in formation of recombinant gametes, i.e. gametes with recombinant genotype Ab and aB.



Effect of different chiasma positions on the separation of the alleles of two linked genes: chiasma has to form at the precise location to disrupt the linkage group.

The **further the gene loci** of the two genes on the same chromosome, **the greater the likelihood of crossing over occurring**. Thus, the **chance of the alleles of the genes being inherited together is lowered**, and **chance of forming recombinant gametes is higher.**

Conversely, if the **gene loci** of linked genes on the same chromosome are **close together**, the **chance of crossing over is lowered**. As a result, the **chance** that the alleles of the genes **will be inherited together** as one linkage group is **higher**, and **less chance of forming recombinant gametes**.

Note: if two gene loci are **completely linked** (i.e. **no crossing over** takes place between the two genes as they are very close to one another), **no recombinants** will result.

Notes to self

possible to have 1:1:1:1 if the 2 genes are very far apart.

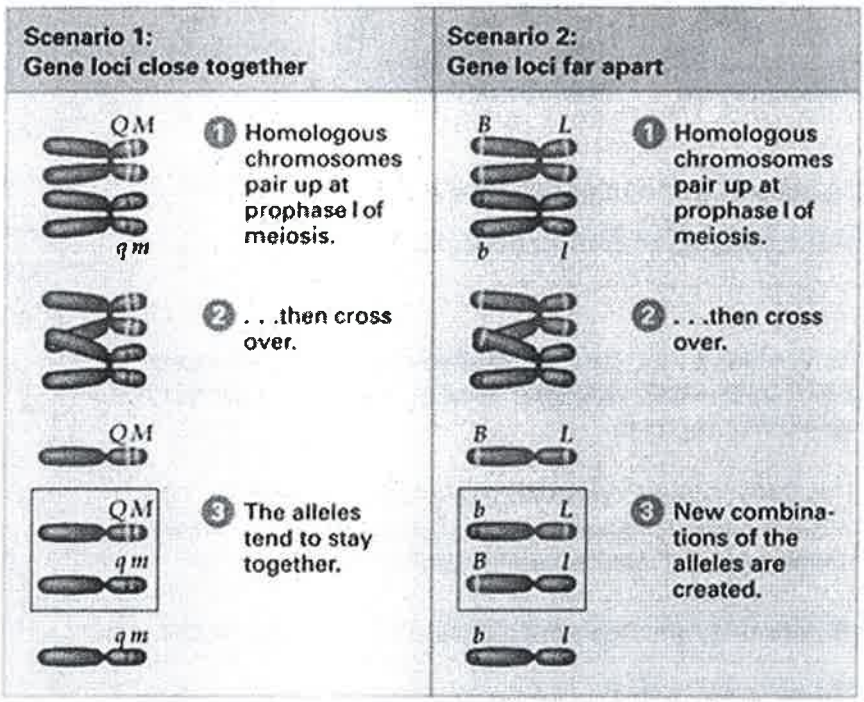
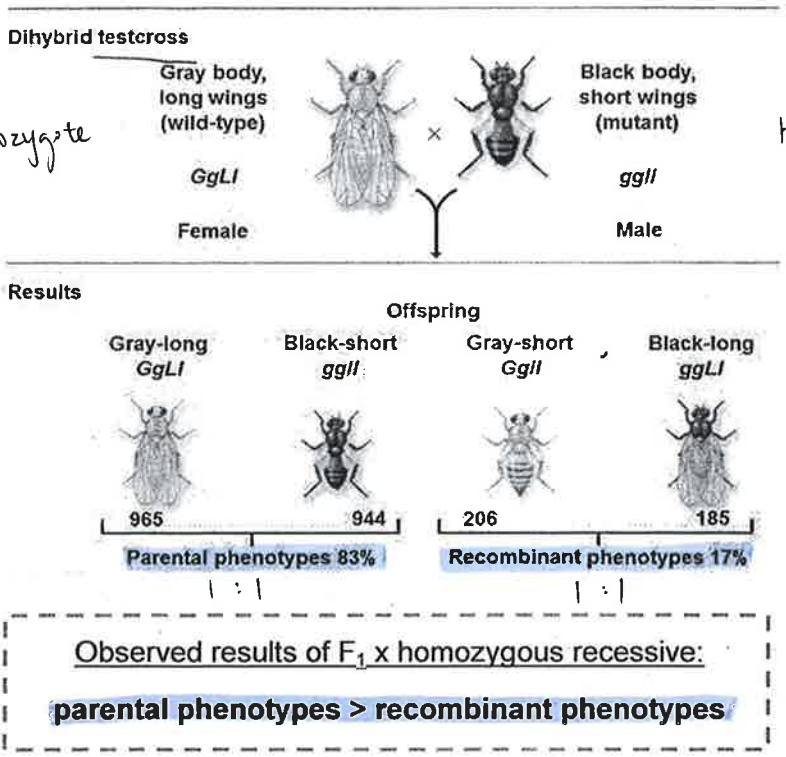


Diagram illustrates the frequency of crossing over & recombination as a result of distance between the two genes on the same chromosome.

T.H. Morgan's experiment of *Drosophila*:

Notes to self



If genes are NOT linked, equal proportions of all 4 types of offspring 1:1:1:1

Observed results of F_1 x homozygous recessive:

parental phenotypes > recombinant phenotypes

- Phenotypes of the parental flies of F_1, i.e. **parental phenotypes** (grey body and normal wings, black body and vestigial wings) are represented in **higher** numbers amongst offspring flies.

  This is due to genes for body colour and wing size being located on the same chromosome; more precisely, alleles G and L are linked in female parental fly of F_1 while alleles g and l are linked in male parental fly of F_1.

  Note: The **two parental phenotypes** are **approximately equal** in numbers.

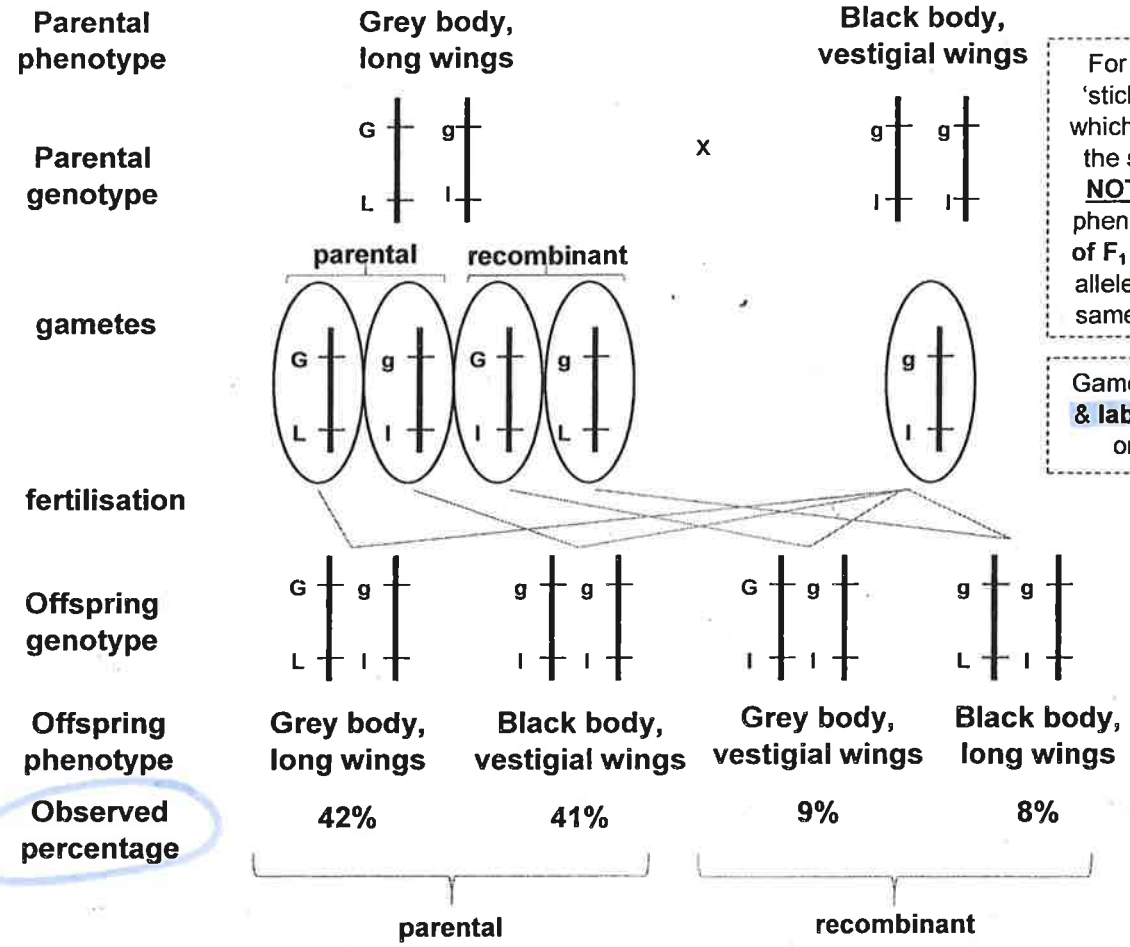
- **Recombinant phenotypes** (grey body and vestigial wings, black body and normal wings) are represented in **lower** numbers amongst offspring flies.

  This is because recombinant gametes are formed as a result of crossing over. As such, the number of recombinants is **dependent on the proximity of the linked genes**. If two genes are close together on a chromosome, the chances of their alleles being separated by crossing over are smaller than if they were far apart. The **further the distance** between two linked genes, the **greater the chance of crossing over**, and thus **higher the percentage of recombinant offspring**.

  Note: The **two recombinant phenotypes** are **approximately equal** in numbers.

- When **heterozygous individual undergoes a test cross**:
  - **expected offspring phenotypic ratio** (i.e. no linkage) = **1** grey body, long wings : **1** black body, vestigial wings : **1** grey body, vestigial wings : **1** black body, normal wings
  - **deviation from this usual Mendelian offspring phenotypic ratio of 1:1:1:1** is indicative of **autosomal linkage**.

Genetic diagram to represent the above results of T.H. Morgan's experiment on *Drosophila*:



For linked genes, draw 'stick' diagrams to show which alleles are found on the same chromosome; **NOTE**: the genotype or phenotype of the **parents of F_1** will determine which alleles are located on the same chromosome in F_1.

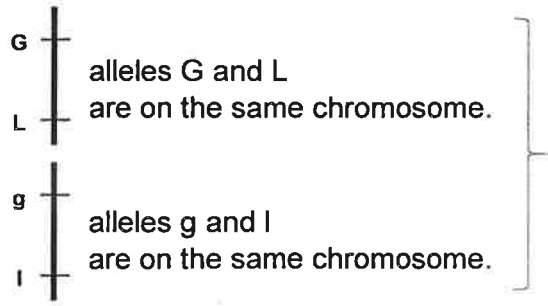
Gametes must be **circled & labelled as "parental" or "recombinant"**

Note: deviation from expected ratio of 1 : 1 : 1 : 1

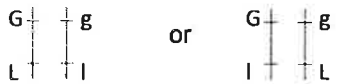
Label offspring as "parental" or "recombinant"

recombination frequency = 9% + 8% = 17%

Results of F_1 test cross:
the larger number of parental phenotypes to recombinant phenotypes indicates that parental genes are linked. More precisely:



NOTE:
the genotype or phenotype of the purebred **parents of F_1** can also be used to determine which alleles are located on the same chromosome in F_1, i.e. whether genotype of F_1 is



Total percentage of recombinant offspring = **recombination frequency**.

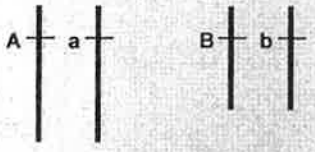
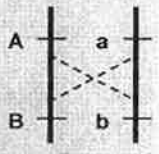
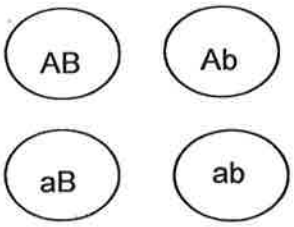
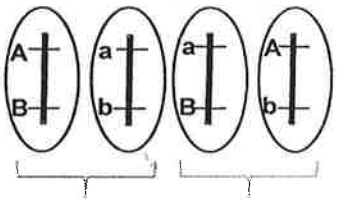
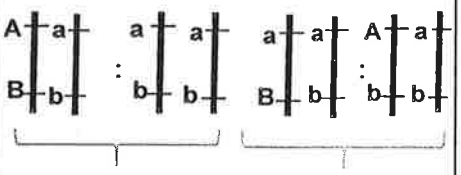
The **distance between two genes** can be **represented in map unit or centimorgan (cM)**:

Recombination frequency of 1 % = 1 map unit or 1 cM

Therefore with a recombination frequency of 17%, distance between the genes for body colour and type of wings in *Drosophila* = 17 map unit or 17 cM

In summary, non-linked genes vs linked genes:

Notes to self

In F_1 individual:	**If genes are on separate chromosomes** (gene locus A/a and gene locus B/b are on different chromosomes) 	**Linked genes** (i.e. gene locus A/a and gene locus B/b are on same chromosome): **crossing over occurs, recombinant gametes formed** 
Gametes of F_1 with genotype AaBb		 parental recombinant
Testcross (F_1 x aabb): ***offspring genotypic ratio***	1AaBb:1Aabb:1aaBb:1aabb	 parental recombinant deviates from usual genotypic ratio of 1:1:1:1
& ***offspring phenotypic ratio***	1 : 1 : 1 : 1	Both parental phenotypes are high in numbers (approximately 1:1), & both recombinant phenotypes are low in numbers (approximately 1:1), deviates from usual phenotypic ratio of 1:1:1:1
(F_1 x F_1): **F_2** ***genotypic ratio*** ***&*** ***F_2 phenotypic ratio***	9A_B_ : 3A_bb : 3aaB_ : 1aabb	deviates from 9:3:3:1
	9 : 3 : 3 : 1	deviates from 9:3:3:1

5. Epistasis

One gene masks the expression of another gene.

Notes to self

Epistasis is a form of gene interaction in which:

- **two or more genes** determine **a single phenotype**
- gene products of different genes are involved in the **same biochemical / metabolic pathway** or **same developmental pathway**
- phenotypic expression of a gene at one locus **masks** the phenotypic expression of a gene at a second locus. The gene whose phenotype is expressed is said to be **epistatic**, while the gene whose phenotype is masked or suppressed is said to be **hypostatic**
  (Note: Epistasis should not be confused with dominance, which is an interaction between alleles at the *same* gene locus.)
- Epistatic alleles can be either dominant or recessive and each type gives a **modified 9:3:3:1** dihybrid **F_2 phenotypic ratio**

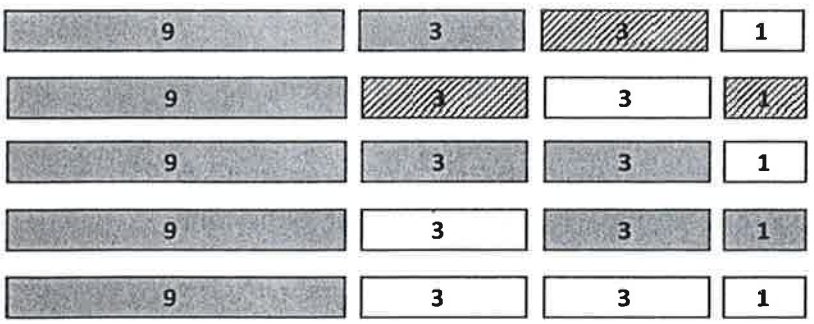
colourless precursor → (functional enzyme A (gene locus 1)) → magenta pigment → (functional enzyme B (gene locus 2)) → blue pigment

Recessive epistasis: → the one that masks is usually upstream
a recessive epistatic allele masks the phenotypic effects of alleles of the other gene when the recessive epistatic allele is present in the homozygous condition.

Dominant epistasis: → usually involves inhibitor
a dominant epistatic allele masks the phenotypic effects of alleles of the other gene when it is present in the homozygous or heterozygous condition.

In epistasis, we will observe **deviation from the expected F_2 phenotypic ratio of 9:3:3:1** when two heterozygous individuals are crossed.

Examples of modified 9:3:3:1 F_2 phenotypic ratios due to epistasis:



(dominant)
(recessive)
(dominant)
(dominant)
(recessive)

Each of the rectangular boxes above represents a particular phenotype. For example, in the epistatic ratio of 12:3:1, there are two grey boxes representing 9 units and 3 units respectively (together for a total of 12 units) for one particular phenotype, one striped box with 3 units for another phenotype and finally one white box with 1 unit for the third phenotype.

Note:
Recessive epistasis results in F_2 phenotypic ratios such as 9:7 and 9:3:4.
Dominant epistasis results in F_2 phenotypic ratios such as 12:3:1, 15:1 and 13:3.

Dihybrid ratios add up to **16**. A ratio of 9:3:3 = 15, is less than 16 and will likely indicate lethal alleles rather than epistasis.

Example 1: Flower colour in sweet pea (9:7 F_2 phenotypic ratio)

Notes to self

Bateson and Punnett crossed two different varieties of white-flowered plants. All the F_1 plants had purple flowers. When plants from the F_1 generation were allowed to self-fertilise, the F_2 generation gave a phenotypic ratio of **9 purple flower plant : 7 white flower plant**. Explain the observed results using a genetic diagram.

***Step 1:* deduce the type of genetic inheritance**

F_2 phenotypic ratio is 9:7, deviates from 9:3:3:1 → *EPISTASIS*

***Step 2:* deduce the biochemical pathway**

Epistasis often occurs when two (or more) different gene products (e.g. enzymes) are part of an enzymatic pathway. In this example a colourless precursor molecule must be acted on by two different enzymes to produce the purple pigment.

Allele C codes for a **functional enzyme C**, which converts the **colourless precursor into a colourless intermediate.** Recessive **allele c'** codes for a **non-functional enzyme C**.

Allele P codes for **functional** enzyme **P**, which converts the **colourless intermediate into the purple pigment**. Recessive **allele p'** codes for a **defective enzyme P**.

If an individual is homozygous for either recessive allele (i.e. **c'c'** or **p'p'**) it will not make any functional enzyme **C** or enzyme **P** respectively. When either one of these functional enzymes is lacking, purple pigment cannot be made and the flower remains white.

Recall: **F_2 genotypic ratio** is **9 C_P_ : 3 C_p'p' : 3 c'c'P_ : 1 c'c'p'p'**

→ **F_2 phenotypic ratio** is **9 purple flower plant : 7 white flower plant**

Note: F_2 phenotypic ratio is deduced with **reference to the biochemical pathway**.

This is an example of **recessive epistasis**.
Genotype c'c' is epistatic to gene locus P/p', and **genotype p'p' is epistatic to gene locus C/c'**.

→ 2 markings involved.

Step 3:

Notes to self

draw genetic diagram to explain how the observed ratio of 9 purple flower plant : 7 white flower plant is obtained

Parental phenotype	White	X	White
Parental genotype	**CCp'p'**		**c'c'PP**
Meiosis, gametes	(Cp')		(c'P)
F_1 genotype		**Cc'Pp'**	
F_1 phenotype		All purple	
Self-cross of F_1 generation		**Cc'Pp' X Cc'Pp'**	
Meiosis, gametes	(CP) (Cp') (c'P) (c'p')		(CP) (Cp') (c'P) (c'p')

Fertilisation

Gametes	(CP)	(Cp')	(c'P)	(c'p')
(CP)	CCPP purple flower plant	CCPp' purple flower plant	Cc'PP purple flower plant	Cc'Pp' purple flower plant
(Cp')	CCPp' purple flower plant	CCp'p' white flower plant	Cc'Pp' purple flower plant	Cc'p'p' white flower plant
(c'P)	Cc'PP purple flower plant	Cc'Pp' purple flower plant	c'c'PP white flower plant	c'c'Pp' white flower plant
(c'p')	Cc'Pp' purple flower plant	Cc'p'p' white flower plant	c'c'Pp' white flower plant	c'c'p'p' white flower plant

F_2 genotypic ratio **9 C_P_ : 3 C_p'p' : 3 c'c'P_ : 1 c'c'p'p'**

F_2 phenotypic ratio 9 purple flower plant : 7 white flower plant

Example 2a: Flower colour in blue-eyed Mary (9:3:4 F_2 phenotypic ratio)

Notes to self

When F_1 plants with blue flowers were allowed to self-fertilise, the F_2 generation gave a phenotypic ratio of **9 blue flower plant : 3 magenta flower plant : 4 white flower plant**. Explain the observed results using a genetic diagram.

***Step 1:* deduce the type of genetic inheritance**

F_2 phenotypic ratio is 9:4:3, deviates from 9:3:3:1 → *EPISTASIS*.

***Step 2:* deduce the biochemical pathway**

Epistasis often occurs when two (or more) different gene products (e.g. enzymes) are part of an enzymatic pathway. In this example a colourless precursor molecule is converted to magenta pigment, which in turn is converted to blue pigment.

Allele M codes for a **functional enzyme M**, which converts the **colourless precursor into a magenta pigment.** The recessive **allele m'** codes for a **non-functional enzyme M**.

Allele B codes for **functional enzyme B**, which converts the **magenta pigment into the blue pigment**. Recessive **allele p'** codes for a **defective enzyme P**.

If an individual is homozygous for recessive allele m' (i.e. **m'm'**), it will not make any functional enzyme M and flower remains **white**.

If an individual is homozygous for recessive allele b (i.e. **bb**) it will not make any functional enzyme **B** and flower is magenta in colour pr.

Recall: **F_2 genotypic ratio = 9 B_M_ : 3 bbM_ : 3 B_m'm' : 1 bbm'm'**

F_2 phenotypic ratio = 9 blue flower plant : 3 magenta flower plant : 4 white flower plant

Note: F_2 phenotypic ratio is deduced with **reference to the biochemical pathway**.

This is an example of **recessive epistasis**.
Genotype m'm' is epistatic to gene locus B/b'.

Step 3:

draw genetic diagram to explain how the observed ratio of 9 blue flower plant : 3 magenta flower plant : 4 white flower plant is obtained

Notes to self

Example 2b:
Coat colour inheritance in Labrador retrievers (9:3:4 F_2 phenotypic ratio)

Coat colour inheritance in Labrador retrievers is determined by two genes. Two **alleles B** and **b**, of a **pigment gene**, results in production of **black** and **brown pigment** respectively. Another gene locus C/c' determines if colour pigment is deposited; allele **C** allows **pigment deposition** in the coat, while with genotype **c'c'**, pigment will not be deposited even if pigment is synthesised and this results in the **gold**/yellow fur phenotype.

Pure bred black Labradors (**CCBB**) were mated with gold Labradors (**c'c'bb**). The offspring were all black. The black Labradors that are heterozygous for both genes (**Cc'Bb**) were mated. What will be the expected phenotype of the F_2 generation? Show your answer in a genetic diagram in the space below.

Step 1: deduce the type of genetic inheritance

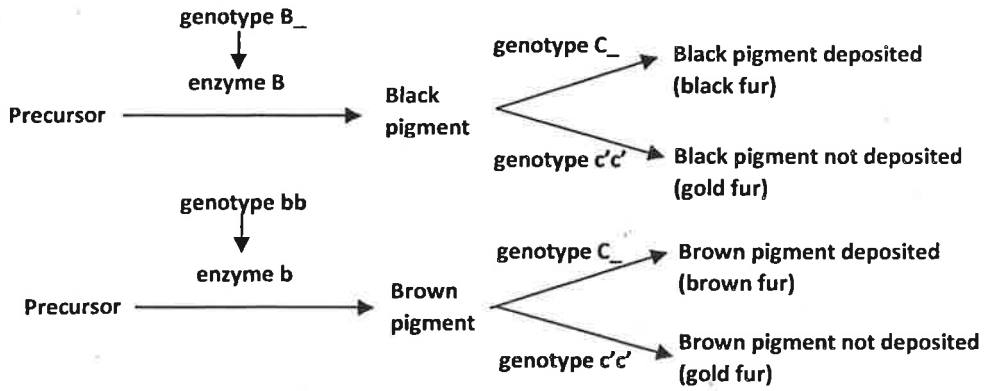
- 2 genes control one phenotype, coat colour
- Genotype c'c' masks the gene locus B/b

} *EPISTASIS*

Note: This case of epistasis is *not* caused by a block in a pathway leading to dark pigment. Gold/yellow dogs can make black or brown pigment, as can be seen in their nose and lips. However, without allele **C**, no black/brown pigment will be deposited in the hairs. In this case, the epistatic gene is "*developmentally downstream*"; the dominant allele **C** must be present before pigment can be deposited.

This is a case of **recessive epistasis**. Genotype **c'c'** is epistatic over the gene locus **B/b**.

Step 2: deduce the developmental pathway



Allele **B** codes for functional enzyme **B** which produces **black** pigment. Allele **b** codes for functional enzyme **b** which produces **brown** pigment. For another separate gene locus C/c, **allele C** allows **colour deposition** in the coat and genotype **c'c'** prevents deposition, resulting in the gold fur phenotype.

Recall: **F_2 genotypic ratio = 9 B_C_ : 3 bbC_ : 3 B_c'c' : 1 bbc'c'**

F_2 phenotypic ratio = 9 black fur : 3 brown fur : 4 gold fur

F_2 phenotypic ratio is deduced with **reference to the developmental pathway**.

Step 3:

draw genetic diagram to explain how the F_2 phenotypic ratio of 9 black fur : 3 brown fur : 4 gold fur is obtained

Example 3: Kernel colour in wheat (15:1 F_2 phenotypic ratio)

Notes to self

A pure line wheat plant with a coloured kernel (genotype = **AABB**) is crossed to plant with white kernels (genotype = **aabb**). All F_1 offspring had coloured kernel. When the resulting F_1 plants were selfed, the offspring gave a ratio of **15 coloured kernels : 1 white kernel**. Explain the observed results using a genetic diagram.

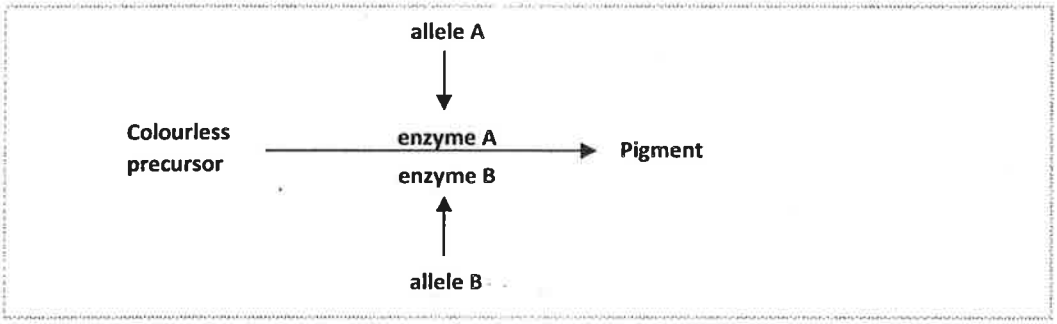
***Step 1:* deduce the type of genetic inheritance**

- 2 genes control one phenotype, kernel colour
- F_2 phenotypic ratio is 15:1, deviates from 9:3:3:1

} *EPISTASIS*

***Step 2:* deduce the biochemical pathway**

For this biochemical pathway, a **functional enzyme A or B** can produce a coloured pigment product from a common colourless precursor. Therefore, only one dominant allele at either of the two gene loci is required to generate the pigment to give colour to the wheat kernel.



Recall: **F_2 genotypic ratio = 9 A_B_ : 3 A_bb : 3 aaB_ : 1 aabb**

F_2 phenotypic ratio = 15 coloured kernel : 1 white kernel

F_2 phenotypic ratio is deduced with **reference to the biochemical pathway**.

This is a case of **dominant epistasis**.

Genotype **A_** is epistatic over the gene locus **B/b** gene locus, and genotype **B_** is epistatic over the gene locus **A/a**.

Step 3:

draw genetic diagram to explain how the observed ratio of 15 coloured kernel : 1 white kernel is obtained

Example 4: Petal colour in *Primula* plant (13:3 F_2 phenotypic ratio)

Notes to self

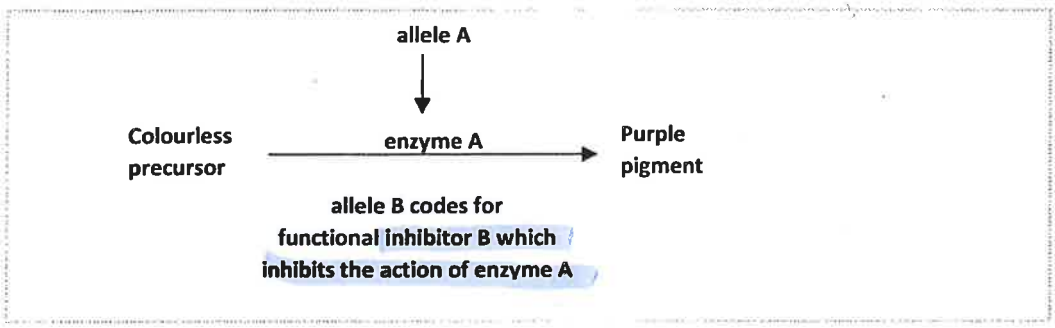
When *Primula* plants with white flowers were allowed to cross-fertilise, a phenotypic ratio of **13 white flower plant : 3 purple flower plant** was obtained in the offspring generation. Explain the observed results using a genetic diagram.

***Step 1:* deduce the type of genetic inheritance**

F_2 phenotypic ratio is ~~15:1~~ 13:3, deviates from 9:3:3:1 → *EPISTASIS*

***Step 2:* deduce the biochemical pathway**

For this biochemical pathway, a **functional enzyme A** converts a colourless precursor to purple pigment while a functional **inhibitor B** will inhibit the action of enzyme A. Therefore in the presence of inhibitor B, the colourless precursor will not be converted to purple pigment and flower remains white.



Recall: **F_2 genotypic ratio = 9 A_B_ : 3 aaB_ : 1 aabb : 3 A_bb**

F_2 phenotypic ratio = 13 white flower plant : 3 purple flower plant

F_2 phenotypic ratio is deduced with **reference to the biochemical pathway**.

This is a case of **dominant epistasis**.

Allele **B** is epistatic over the gene locus **A/a** gene locus.

Step 3:

draw genetic diagram to explain how the observed ratio of 13 white flower plant : 3 purple flower plant is obtained

Notes to self

Example 5: Fruit colour in squash (12:3:1 F_2 phenotypic ratio)

In summer squash, fruit colour may be white, yellow or green. White fruits are produced in the presence of allele W. At a second gene locus, allele G results in yellow fruits, while allele g results in green fruits. When two squash plants with white fruit were crossed, a phenotypic ratio of **12 white squash : 3 yellow squash : 1 ~~white~~ green squash** was obtained in the offspring generation. Draw a genetic diagram to observed results.

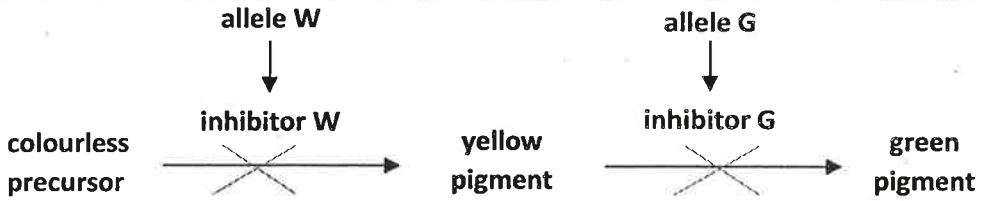
→ pathway will not proceed

***Step 1:* deduce the type of genetic inheritance**

- 2 genes control one phenotype, squash colour
- F_2 phenotypic ratio is 12:3:1, deviates from 9:3:3:1 — *EPISTASIS*

***Step 2:* deduce the biochemical pathway**

For this biochemical pathway, alleles W and G code for **functional inhibitors W** and **G** respectively. Inhibitor W inhibits the enzyme that converts the colourless precursor to yellow pigment. As such, in the presence of inhibitor W, squash remains white. Inhibitor G inhibits the enzyme that converts the yellow pigment to green pigment. In the presence of inhibitor G, the squash is yellow if functional inhibitor W is not present.



Recall: **F_2 genotypic ratio = 9 W_G_ : 3 W_gg : 3 w'w'G_ : 1 w'w'gg**

F_2 phenotypic ratio = 12 white squash : 3 yellow squash : 1 green squash

F_2 phenotypic ratio is deduced with **reference to the biochemical pathway**.

This is a case of **dominant epistasis**.

Allele **W** is epistatic over the gene locus **G/g** gene locus.

Step 3:
draw genetic diagram to explain how the observed ratio of 12 white squash : 3 yellow squash : 1 green squash is obtained

6. Chi-squared test

Notes to self

The chi-square test is used when a set of **observed results** or **observed ratio** needs to be **compared** to a set of **expected results** or **a known ratio**.

For instance, according to Mendel's laws, a 3: 1 offspring phenotypic ratio is expected (e.g. 3 purple flower plants : 1 white-flower plant) when two heterozygotes undergo a monohybrid cross. However, the actual observed numbers for the offspring of such a cross were 65 purple-flowered plants and 35 white-flowered plants. In such cases:

- What is the "goodness to fit" between the observed and expected results?
- Were the deviations (differences between observed and expected) the result of chance alone, or were they due to other factors?
- How much deviation can occur before we can conclude that **something other than chance** is at work, causing the observed to differ from the expected?

By carrying out the chi-square test we can make one of the following conclusions:

Either

- the difference between the observed and expected results is not significant, and any difference is due to chance alone (i.e. H_0 is not rejected)

or

- the difference between the observed and expected results is ~~not~~ significant, and any difference is due to other factors and not chance alone (i.e. H_0 is ~~not~~ rejected)

Therefore as a student, you must know how to:

- carry out a chi-square test (which involves 5 steps), and
- determine whether to reject H_0 or not to reject H_0.

  (H_0 = difference between the observed and expected results is not significant, and any difference is due to chance alone)

You will be given a chi-square table and the following chi-square formula in the exams. You must know how to use the formula properly, i.e. carry out the correct mathematical operations denoted by the symbols in the formula. You will also need to know how to calculate the degree of freedom in order to use the chi-square table.

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

This is the chi-square formula which you need to use to calculate the chi-square value (χ^2).

Σ = summation, **O** = observed value, **E** = expected value

$$df = n - 1$$

You will also need to calculate the degrees of freedom (***df***),

where ***n*** = the number of phenotype classes.

probability of H_0 being true.

STEPS in carrying out a chi-square test:

Notes to self

Step 1: Determine the expected offspring phenotypic ratio.

Step 2: Calculate the χ^2 value.

Step 3: Determine the degrees of freedom (*df*).

Step 4: Determine "p", using the *df* and χ^2 value and chi-square table.

Step 5: Reject or do not reject H_0 based on 5% significance level. Draw your conclusion.

Example:

A homozygous white flowered, long-stemmed tobacco plant, *Nicotiana affinis* was crossed with a homozygous pink flowered, short-stemmed plant. The F_1 generation had white flowers and long stems. When the F_1 generation was crossed with recessive tester stock, the following progeny were obtained:

51 white flowered, long-stemmed plants,

46 white flowered, short-stemmed plants,

49 pink flowered, long-stemmed plants,

44 pinked flowered, short-stemmed plants,

Do the results indicate that the inheritance of the genes for flower colour and stem length follow Mendel's laws? Explain, using χ^2–test to support your answer.

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

Where

O = observed value;

E = expected value;

$\sum$ = sum of

Distribution of χ^2

degrees of freedom	*probability, p*				
	0.10	0.05	0.02	0.01	0.001
1	2.71	3.84	5.41	6.64	10.83
2	4.61	5.99	7.82	9.21	13.82
3	6.25	7.82	9.84	11.35	16.27
4	7.78	9.49	11.67	13.28	18.47

Step 1: Determine the expected offspring phenotypic ratio

The expected offspring phenotype ratio is:

1 white flowered, long-stemmed plant : **1** white flowered, short-stemmed plant : **1** pink flowered, long-stemmed plant : **1** pink flowered, short-stemmed plant

Step 2: **Calculate the χ^2 value**

Notes to self

Total number of offspring = 51 + 46 + 49 + 44 = 190

Expected frequency = 190 ÷ 4 = **47.5**

(since we expect equal numbers of offspring for each class of phenotype)

offspring phenotype	Observed frequency (O)	Expected frequency (E)	$(O-E)^2 / E$
white flowered, long-stemmed plants	51	47.5	$(51-47.5)^2 \div 47.5 = 0.258$
white flowered, short-stemmed plants	46	47.5	$(46-47.5)^2 \div 47.5 = 0.0474$
pink flowered, long-stemmed plants	49	47.5	$(49-47.5)^2 \div 47.5 = 0.0474$
pinked flowered, short-stemmed plants	44	47.5	$(44-47.5)^2 \div 47.5 = 0.258$

$\chi^2 = 0.61$

Step 3: **Determine the degrees of freedom (*df*)**

***df* = n – 1 = 4 – 1 = 3** *(where n = number of classes)*

Step 4: **Determine "p", using the *df* and χ^2 value to check against the chi-square table**

With df = 3 and χ^2 value of 0.61, **p > 0.1**

Step 5: **Reject or do not reject H_0 based on 5% significance level. Draw your conclusion.**

Since **p > 0.05** at **5% significance level, do not reject H_0.**

(The probability is very high that any difference is due to chance)

There is **no significant difference** between the observed and expected results. Any difference is due to **chance alone.**

(Note: Lack of evidence against a hypothesis is not evidence for the hypothesis being true. Therefore **we cannot say that we accept H_0.** We can only conclude that there is not have enough evidence to reject the null hypothesis – i.e. we do not reject H_0).

On the contrary, if **p < 0.05** at 5% significance level:

- **H_0 is rejected**. The **difference between observed and expected results is significant** and **is due to other factors other than chance alone.**
- Then the results indicate that the inheritance of the genes for flower colour and stem length do not follow Mendel's laws. Some other factor could have contributed to the difference between the observed and expected values.

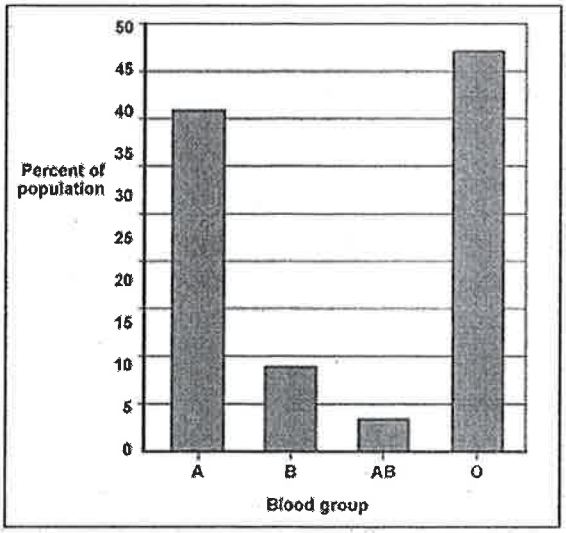
7. Discontinuous Variation & Continuous Variation

Notes to self

a. Discontinuous Variation

Features:

- There are **distinct** and **discrete** phenotypes.
- Generally caused by **different alleles** of a **single gene** or a few genes.
- Their phenotypic expression in many cases are **unaffected by environmental conditions.**
- e.g. blood groups in the ABO system - A, B, AB or O; green and yellow peas; wrinkled and smooth peas.



Discontinuous (discrete) variation showing the percentage of individuals in a population with four different (discrete) blood types.

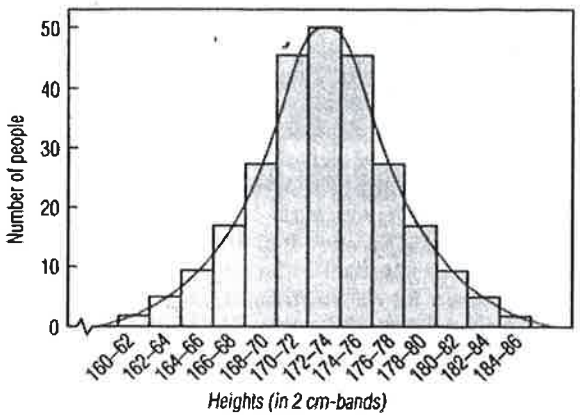
b. Continuous Variation

Notes to self

Features:

①
- Differences are slight and **phenotypic differences vary along a continuum** in gradations. **No distinct groupings** are formed.
- There is a **range of phenotypes for a characteristic** e.g. human exhibit continuous variation in height, weight and skin colour.

continuous variation variation in the height of adult male humans; the results cluster quite closely around one value, and show a normal distribution (in this type of distribution the mean, median and mode values coincide); for convenience in plotting, the heights are grouped into 13 arbitrary groups, each covering a height range of 2 cm



Example of continuous variation - height of adult human males.

②
- Continuous variation usually is the combined effect of **many genes** and therefore indicates **polygenic inheritance.** There is an **additive effect** of **two or more** (usually many) **genes** on a single phenotypic character.
- Polygenic inheritance will result in a **bell-shape normal distribution** curve (unbroken range of phenotypes), where **intermediate phenotypes are more common than extreme phenotypes.**

③
- **Phenotypes are affected by environmental factors.** For example, exposure to sun, also affects the skin-colour phenotype and make the graph smooth rather than a step-like histogram.

Notes to self

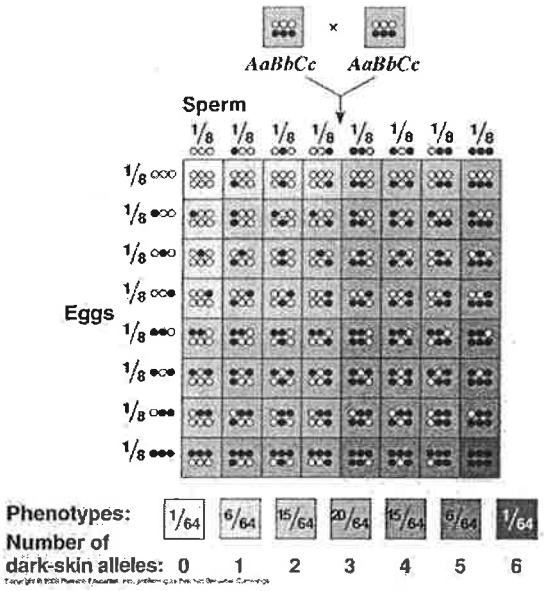


Figure A

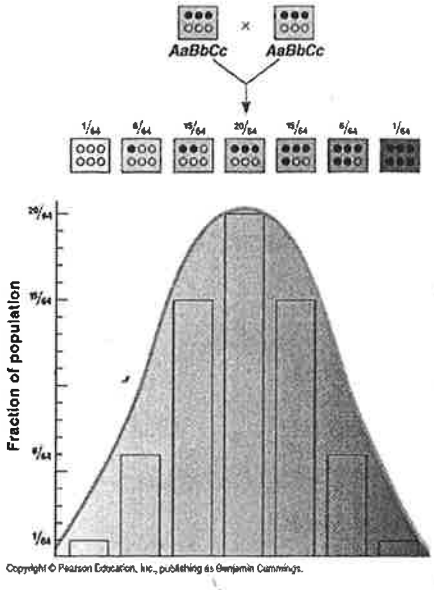


Figure B

Figure A

A simplified model for the polygenic inheritance of skin colour.

According to this model three separately inherited genes affect the darkness of skin.

The heterozygous individuals (**AaBbCc**) represented by the two rectangles at the top of these figures carry three dark-skin alleles (black circles, which represent alleles **A**, **B**, or **C**) and three light-skin alleles (white circles which represent alleles **a**, **b**, or **c**).

A dark-skin allele for each gene (**A, B,** or **C**) contributes one "unit" of darkness to the phenotype. An individual **AABBCC** would be **very dark**, while an **aabbcc** individual would be **very light**. An **AaBbCc** person would have **intermediate shade**.

Because the alleles have a cumulative effect, the genotype **AaBbCc** and **AABbcc** would make the same genetic contribution (three units) to skin darkness.

The Punnett square shows all the possible genetic combinations in the gametes and the resulting offspring of a large number of hypothetical matings between these heterozygotes. The results are summarized by the phenotype ratios under the Punnett square.

Figure B

Represents a bar chart of the results, with skin colour (number of dark-skin alleles) along the x-axis and fraction of offspring along the y-axis.