

What I need to know ...01

- 2(k) Explain the terms locus, allele, dominant, recessive, codominant, incomplete dominance, homozygous, heterozygous, phenotype, genotype and sex linkage
- 2(I) <u>Explain</u> how genes are inherited from one generation to the next via the germ cells or gametes
- 2(n) <u>Use genetic diagrams</u> to solve problems in dihybrid crosses



- Common understanding of the term
- Allows scientists, researchers and doctors to be on the "same page" during discussions
- Important in solving genetic crosses

02

.[2]

- 2011 STQ Paper
 - (a) Explain what is meant by the terms dominant and allele.

dominant	•••••••••••••••••••••••••••••••••••••••
•••••••••••••••••••••••••••••••••••••••	
allele	

 $\mathbf{02}$

- 2015 STQ Paper
 - (d) The inheritance of sickle cell anaemia is autosomal recessive.

Explain what is meant by autosomal recessive inheritance.

.....[2]

02

List of words

- Locus
- Allele (Dominant & Recessive)
- Codominant
- Incomplete dominant
- Homozygous & Heterozygous
- Phenotype
- Genotype
- Sex Linkage

 $\mathbf{02}$

Locus (singular)

• Loci (plural)

Pg.1

 The fixed position on a particular chromosome that each gene occupies







02

List of words

✓ Locus

- Allele (Dominant & Recessive)
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Definitions

02

<u>Allele</u>

- Alternative form of a gene
- Occupy the same gene locus on homologous chromosomes
- Occur in pairs in a diploid cell
- Causes slightly different phenotypes for the same characteristic
- Due to a slight difference in DNA seq.





Definitions



Dominant allele Gene is transcribed into mRNA, and mRNA being translated into polypeptide

Allele whose effect is expressed in the phenotype in <u>both</u> homozygous and heterozygous conditions





Recessive allele

Pg.1

Gene is transcribed into mRNA, and mRNA being translated into polypeptide

 Allele whose effect is expressed in the phenotype <u>only</u> in the presence of another identical allele / in homozygous condition



Definitions

Codominant

- Both alleles are <u>equally expressed</u>
- in the phenotype of the heterozygote



Hb^AHb^A

Normal



Hb^AHb^S

Sickle cell trait



02

Hb^sHb^s

Sickle cell anaemia

Codominance

White Bull

Red Cow







Roan Offspring (red and white hairs are expressed equally)



02

Incomplete dominance

Pg.1

 Situation where the heterozygote exhibits a phenotype which is <u>intermediate</u> between the homozygous dominant & recessive forms

Generally due to the **quantitative effect** of the number of copies of a wild-type allele

Two copies produce the **most** mRNA transcripts & functional protein products; **One** copy => less protein products **Zero** copy => no functional transcript/product



02

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Homozygous

Pg.1

- Genotype of two identical alleles of a gene
- at a particular gene locus of homologous chromosomes
- e.g. 2 dominant alleles or 2 recessive alleles in the genotype





Heterozygous

Pg.1

- Genotype of two different alleles of a gene
- at a particular gene locus of homologous chromosomes





02

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Test Yourself!

Which of the following statements about recessive alleles is correct?

- 1. They are expressed phenotypically when homozygous.
- 2. They are not expressed phenotypically when codominant.
- 3. They are masked by dominant alleles in homozygotes.
- 4. They do not undergo transcription in heterozygotes.

Definitions



Genotype :

 Is determined by the alleles of specific genes / genetic make-up with respect to the alleles





Phenotype :

Pg.2

 The physical manifestation / trait resulting from a specific genotype





Phenotype :

Pg.2

 The physical manifestation / trait resulting from a specific genotype



Definitions



Phenotype :

- The physical manifestation / trait resulting from a specific genotype
- And its interaction with the environment





Extra information...

• Explain how genotype is linked to phenotype

01



page 15

Alleles

on DNA

transcription

mRNA

translation

protein

interactions

phenotype

How genotype is linked to phenotype 05

- Diff. alleles differ slightly in their DNA sequences
 - Diff. alleles **transcribed** to form diff. **mRNA**;
 - Diff. mRNA translated into diff.
 polypeptide chain(s) / protein(s);

[Different alleles will result in the production of different polypeptide chain(s) / protein(s)]

- Different gene products / proteins may affect different cellular pathways / different parts of a pathway / different functions;
- Resulting in **different phenotypes**

How genotype is linked to phenotype

[At gene gene loci > diff. genotype > diff. phenotype]

- The alleles present for each particular gene may be dominant or recessive;
- Dominant alleles mask the effect / influence of recessive alleles;
- Hence only the effect of the dominant allele will be expressed in the phenotypes of homozygous dominant and heterozygous organisms;
- Phenotypes of recessive individuals will only be expressed when both alleles are recessive

02

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- Phenotype
- Genotype
- Sex Linkage

Definitions



Sex Linkage:

- Situation where a gene is located on a sex chromosome.
- This results in the characteristic being expressed mainly in one sex.



02

List of words

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- Homozygous & Heterozygous
- Codominant
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- Phenotype
- Genotype
- 🗸 Sex Linkage

(Additional) Definitions 02

• Gene

- Heritable sequence of nucleotides along a DNA molecule which codes for a polypeptide and hence may result in a phenotypic effect.

Gene pair

- Two alleles of a particular gene present in a diploid cell



(Additional) Definitions

 $\mathbf{02}$

• Cross

- Deliberate mating of 2 individuals in genetic analysis

Selfing

- Fertilisation of female gamete by a male gamete from the same individual. Used for plants

Sibling mating

- Mating between siblings.

Dangerous due to inbreeding. Offspring have a high chance of inheriting any deleterious alleles in the family gene pool.

Because of this, plants practice fruit and seed dispersal whereas animals practice sex-biased dispersal, kin avoidance and kin selection.
(Additional) Definitions

- Pure line / true-breeding / pure breeding
- Selfing over many generations can produce a pure line

 $\mathbf{02}$

- Members of a pure line are homozygous at each gene locus.
- They are said to breed true or are true-bred or pure bred
- They are either homozygous dominant or homozygous recessive but never heterozygous





Gregor Johann Mendel 1822 - 1884



1822-1884; Czech Republic; family of farmers

Priest / teacher in monastery

Dreams

Scientist as well

learning

What organisms did he research on?

GREGOR MENDEL

Educational background?

Thanks to

work in g

eas and me

"Are there invisible "factors"—now called genes—in determining the traits of an organism???"

matter how many times they try.

1822-1884; Czech Republic; family of farmers

Priest / teacher in monastery

Scientist as

well

Dreams

Joy of

learnir

GREGOR MENDEL

Educational background?

"Are there invisible "factors"—now called genes—in determining the traits of an organism???"

w many times they try.

Say "NO" to study of animal sex

Mendel's 2 Laws

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- Gregor Mendel observed the patterns of inheritance in pea plants and formulated two laws in the 1860s.
- Both laws could be related to the behaviour of chromosomes during meiosis.
- 1. Law of Segregation

Pg.4

2. Law of Independent Assortment

Pg.4 Law of Segregation 03



- Involves a single gene locus only (gene A)
- Related to the separation of homologous chromosomes during anaphase I of meiosis

Pg.4 Law of Segregation **03**



The two alleles (alleles A and a) for each characteristic segregate during anaphase I of meiosis, which occurs during the formation of gametes.

Pg.4 Law of Segregation **03**



- Half of the gametes carry one allele of a gene pair while the other half carries the other allele.
- The Law of segregation was based on work involving <u>monohybrid crosses</u>.

i.e. the study of inheritance of **one** trait controlled by **one** gene

Law of Segregation 03





A pure-breeding tall pea plant was crossed with a purebreeding short pea plant. Both are referred to as the parental generation.





A pure-breeding tall pea plant was crossed with a pure-breeding short pea plant. Both are referred to as the parental generation.

<u>Key :</u>

Let T represent the allele for tall plant

Let t represents the allele for short plant

A key shows

- symbols of alleles used
- relationship between alleles

The allele for tall plant (T) is dominant to the allele for short plant (t)



Provide a key if not given in question.

Choose an alphabet to represent the trait (height of plant) Use the **capital letter** of the chosen alphabet to represent the **dominant allele** for the trait. Dominant allele accounts for the dominant (tallness) phenotype.

Use the **small letter** of the chosen alphabet to represent the **recessive allele** for the trait. Recessive allele accounts for the recessive (shortness) phenotype.



- Parental phenotype
- Parental genotype
- Parental gametes
- F1 genotype
- F1 phenotype

- Tall plantShort plantTTXttTGametes are always circled!tTtTtTt
 - All tall plants



2





What will be the results of selfing the F1 plants?

How to use a Punnett square

How to use a Punnett square





Selfing of F1 (self-pollination)





Selfing of F1 (self-pollination)

Parental phenotype Parental genotype

Parental gametes

Punnett square



Law of Segregation 03





Selfing of F1 (self-pollination)



3 tall plants F2 phenotypic ratio 2

1 short plant



A heterozygous tall pea plant was crossed with a purebreeding short pea plant.

Both are referred to as the parental generation. Tall trait is dominant over short trait.

What will be the genotypes, phenotypes and phenotypic ratio of the F_2 generation after cross fertilization of both F_1 genotypes?



A heterozygote tall pea plant was crossed with a purebreeding short pea plant. Tall trait is dominant over short trait. What will be the genotypes, phenotypes and phenotypic ratio of the F_2 generation <u>after cross fertilization of both F_1 </u> <u>genotypes</u>?

Key :

Let T represent the allele for tall plant Let t represents the allele for short plant The allele for tall plant (T) is dominant to the allele for short plant (t)



Crossing of parental generation

Parental phenotype	:	Tall plant		Short plant
Parental genotype	:	Tt	X	tt
Parental gametes	:	T t		t
F1 genotype	:	Tt		tt
F1 phenotype	:	Tall plant		Short plant
F1 phenotypic ratio	:	1	:	1



Cross fertilisation of both F1 genotypes

F1I phenotype	:	Tall plant		Short plant
F1 genotype	:	Tt	X	tt
F1 gametes	:	$(\mathbf{T}) (\mathbf{t})$		t
F2 genotype	:	Tt		tt
F2 phenotype	:	Tall plant		Short plant
F2 phenotypic ratio	:	1	:	1



Mendel's 2 Laws

UR



- Both laws could be related to the behaviour of chromosomes during meiosis.
- 1. Law of Segregation

Pg.8

2. Law of Independent Assortment

 Mendel's law of independent assortment addresses the process of how two different genes on different pairs of homologous chromosomes segregate



Pg.8

Chromosome 6 Chromosome 3

Gene for colour of seed Gene for smoothness of seed ====→ dihybrid cross



Pg.8

 Orientation of homologous chromosomes shown in

 (a) is as likely to occur as that in (b) because the arrangement of one homologous pair is independent of the other





 Related to the independent arrangement during <u>metaphase I</u>, and subsequent separation of homologous chromosomes during anaphase I.





 The Law of independent assortment was based on work involving <u>dihybrid</u>

<u>crosses</u>

Pg.8

i.e. the study of inheritance oftwo traits controlled bytwo genes



03



Method of Mendel's experiment

- Pure-bred pea plants grown from <u>round seeds with</u> <u>yellow cotyledons</u> were crossed with pure-bred pea plants grown from <u>wrinkled seeds with green</u> <u>cotyledons</u>.
- The above cross gave rise to the F_1 generation.
- The F_1 plants were selfed to produce the F_2 generation.



Dihybrid cross





Method of Mendel's experiment

 Pure-bred pea plants grown from <u>round seeds with</u> <u>yellow cotyledons</u> were crossed with pure-bred pea plants grown from <u>wrinkled seeds with green</u> <u>cotyledons</u>.



2 genes code for 2 traits:1. Shape of seed (round or wrinkled)2. Colour of cotyledon (yellow or green)



Method of Mendel's experiment

- The above cross gave rise to the F₁ generation.
- The F_1 plants were selfed to produce the F_2 generation.


Worked Example (Pg. 10)

Parental: round seeds with yellow cotyledons X wrinkled seeds with green cotyledons F1 generation Which alleles are dominant, and which are recessive? F2 generation



 All individuals of the F₁ generation produced round seeds with yellow cotyledons.



Alleles for round seeds and yellow cotyledons are dominant



Worked Example (Pg. 10)

Results of Mendel's experiment

- All individuals of the F₁ generation produced round seeds with yellow cotyledons.
- The F2 generation showed the following phenotypic ratio



<u>Key :</u>

Let R represent the allele for round seed

- Let r represent the allele for wrinkled seed
- Let Y represent the allele for yellow cotyledon
- Let y represent the allele for green cotyledon

The allele for round seed (R) is dominant to the allele for wrinkled seed (r) The allele for yellow cotyledon (Y) is dominant to the allele for green cotyledon(y).

Parental phenotype	•	Round seed Yellow cotyledon		Wrinkled seed Green cotyledon
Parental genotype	-	RRYY	X	rryy
Parental gametes		RY		ry

Parental phenotype	•	Round seed Yellow cotyledon		Wrinkled seed Green cotyledon
Parental genotype	-	RRYY	Х	rryy
Parental gametes		RY		ry

F1 genotype	•	RrYy
F1 phenotype		All round seed
		yellow cotyledon

Selfing F1 generation									
F1 genotype	-	RrYy		Х	RrYy				
F1 gametes		RY	Ry		RY	Ry			
		rY	ry		rY	ry			

Selfing F1 generation						
F1 genotype		RrYy		X	RrYy	
F1 gametes	•	RY	Ry		RY	Ry
		rY	ry		rY	ry

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
	(Round,yellow)	(Round,yellow)	(Round,yellow)	(Round,yellow)
Ry	RRYy	RRyy	RrYy	Rryy
	(Round,yellow)	(Round,green)	(Round,yellow)	(Round,green)
rY	RrYY	RrYy	rrYY	rrYy
	(Round,yellow)	(Round,yellow)	(wrinkled,yellow)	(wrinkled,yellow)
ry	RrYy	Rryy	rrYy	rryy
	(Round,yellow)	(Round,green)	(wrinkled,yellow)	(wrinkled _g reen)

Selfing F1 generation					
F1 genotype	RrYy		X	RrYy	
F1 gametes	RY	Ry		RY	Ry
	rY	ry		rY	ry



	RY		Ry		r	Y		ry
RY	RRYY (Round,yellov	v)	RRYy (Round,yellow)		RrYY (Round,yellow)		(R	RrYy ound,yellow)
Ry	RRYy (Round,yellov	v)	RRyy (Round,green) (Round,ye		Yy ,yellow)	(R	Rryy Round,green)	
rY	RrYY (Round,yellov	v)	RrYy (Round,yellow) (wr		rrYY (wrinkled,yellow)		(wr	rrYy inkled,vellow)
ry	RrYy (Round,yellov	v)	Rryy (Round,gree	y rrYy reen) (wrinkled,yellow;		(wr	rryy rinkled,green)	
F2 pheno	type	:	Round seed Yellow cotyledon	F	Round Wrinkle seed seed Green Yellow cotyledon cotyledo		ed / on	Wrinkled seed Green cotyledon
F2 pheno	typic ratio	:	9:		3:	3 :		1



Round seed, yellow cotyledon Round seed, green cotyledon Wrinkled seed, yellow cotyledon Wrinkled seed, green cotyledon

Key:

DO NOT USE HIGHLIGHTER OR SHADING IN PUNNETT SQUARE/LEGEND

Law of Independent Assortment 03

- When Mendel considered dihybrid cross, as opposed to monohybrid, resulting F₂ generation (after selfing of F₁) did not have 3:1 dominant : recessive phenotypic ratios.
- The two traits, if considered to inherit independently, fit the principle of segregation.

Pg.11

 Instead of 4 possible genotypes from a monohybrid cross, dihybrid crosses have as many as <u>16 possible genotypes</u>, with phenotypic ratios of <u>9:3:3:1</u>



Sample mark scheme



- Parental gametes (circled)
- F1 genotypes
- F1 phenotypes
- F1 gametes (circled)
- Punnett square showing all F2 genotypes
- Genotypes correspond to phenotypes
 / legends for Punnett square
- F2 phenotypic ratio





In a certain species of a garden plant, pure-bred plants with red flowers and thorny stems were crossed with pure-bred plants with white flowers and smooth stems. This cross gave rise to the F_1 generation. The F_1 plants were selfed to produce the F_2 generation. All the progeny plants of the F_1 generation had red flowers and thorny stems.

5 minutes

Draw a genetic diagram of the cross, including gametes, showing the genotypes and phenotypes of the F_1 and F_2 generations. Give the ratio of phenotypes expected in the F_2 generation.

<u>Key :</u>

Let R represent the allele for red flowers

- Let r represent the allele for white flowers
- Let T represent the allele for thorny stems
- Let t represents the allele for smooth stems
- The allele for red flowers (R) is dominant to the allele for white flowers (r)
- The allele for thorny stems (T) is dominant to the allele for smooth stems (t)

Parental phenotype		Red flowers Thorny stems		White flowers Smooth stems
Parental genotype	-	RRTT	X	rrtt
Parental gametes		RT		rt

Parental phenotype		Red flowers Thorny stems		White flowers Smooth stems
Parental genotype	-	RRTT	Х	rrtt
Parental gametes	-	RT		rt

F1 genotype	RrTt
F1 phenotype	All red flowers, thorny stems

Selfing F1 generation									
F1 genotype		RrTt		X	RrTt				
F1 gametes		RT	Rt		RT	Rt			
		T	rt		rT	rt			

Selfing F1 generation											
F1 genotype		Rr	Tt	X	RrTt						
F1 gametes		RT	Rt		RT	Rt					
		rT	rt		T	rt					



	RT		Rt		rT		rt	
RT	RRTT (Red, thorny)		RRTt (Red, thorny	/)	Rr (Red, f	TT thorny)	RrTt (Red, thorny)	
Rt	RRTt (Red, thorny)		RRtt (Red,smooth	า)	RrTt (Red, thorny)		Rrtt (Red,smooth)	
rT	RrTT (Red, thorny)		RrTt (Red, thorny)		rrTT (white,thorny)		rrTt (white.thorny)	
rt	RrTt (Red, thorny)		Rrtt (Red,smooth)		rrTt (white,thorny)		rrtt (white, smooth)	
F2 phenotype		:	Red flowers Thorny stems	Red flowers Smooth stems		White flowers Thorny stems		White flowers Smooth stems
F2 phenotypic ratio :		9:		3 :	3 :		1	

What I need to know ...01

- 2(k) Explain the terms locus, allele, dominant, recessive, codominant, incomplete dominance, homozygous, heterozygous, phenotype, genotype and sex linkage
- 2(I) <u>Explain</u> how genes are inherited from one generation to the next via the germ cells or gametes
- 2(n) <u>Use genetic diagrams</u> to solve problems in dihybrid crosses

How genes are inherited from one generation 05 and passed to the next via gametes

- Genes are present on chromosomes ;
- during meiosis, chromosomes randomly align at the metaphase plate during metaphase I and are subsequently separated during anaphase I;
- giving rise to haploid gametes ;
- which contain unique combinations of alleles;
- random fusion of 2 gametes gives rise to an offspring with inherited alleles from parents



Scenario	Phenotypic ratio
Monohybrid cross Heterozygote x Heterozygote e.g. Aa x Aa	3:1
Dihybrid cross Heterozygote x Heterozygote	9:3:3:1

e.g. AaBb x AaBb

Pg.13