



2020
Version

With 2019 NCEA
Exam included

Science AS 90948

S1.9 Genetic Variation



Part One

Achievement Criteria (Part ONE)



AS 90948
S1.9

Biological concepts and processes relating to the inheritable nature of DNA will be selected from:

- ☐ the roles of DNA in both carrying instructions to the next generation and determining phenotype
- ☐ the relationship between DNA, alleles, genes, and chromosomes
- ☐ the way in which genotype determines phenotype
- ☐ the way chromosomes exist as pairs so that individuals inherit two copies of each gene.

Biological concepts and processes relating to variation in phenotype will be selected from:

- ☐ the significance of an allele as an alternative version of a gene
- ☐ the role of mutations in forming new alleles
- ☐ the role of meiosis in generating gametes (students are not required to provide the names of the stages of meiosis)
- ☐ the significance of sexual reproduction (in producing a new mix of alleles)
- ☐ the patterns of inheritance involving simple monohybrid inheritance showing complete dominance, sex determination, possible genotypes, and phenotype ratios.

Achievement Criteria (Part TWO)

AS 90948
\$1.9

Biological concepts and processes relating to variation in phenotypes as adaptive features will be selected from:

- ☐ inheritable and non-inheritable variations that exist within a group of living organisms
- ☐ differing rates of survival by various members of a group may depend on their phenotype
- ☐ the importance of variation within populations (population and species survival) in a changing environment such as pest infestation, disease, drought, or flood
- ☐ the advantages and disadvantages of sexual reproduction.

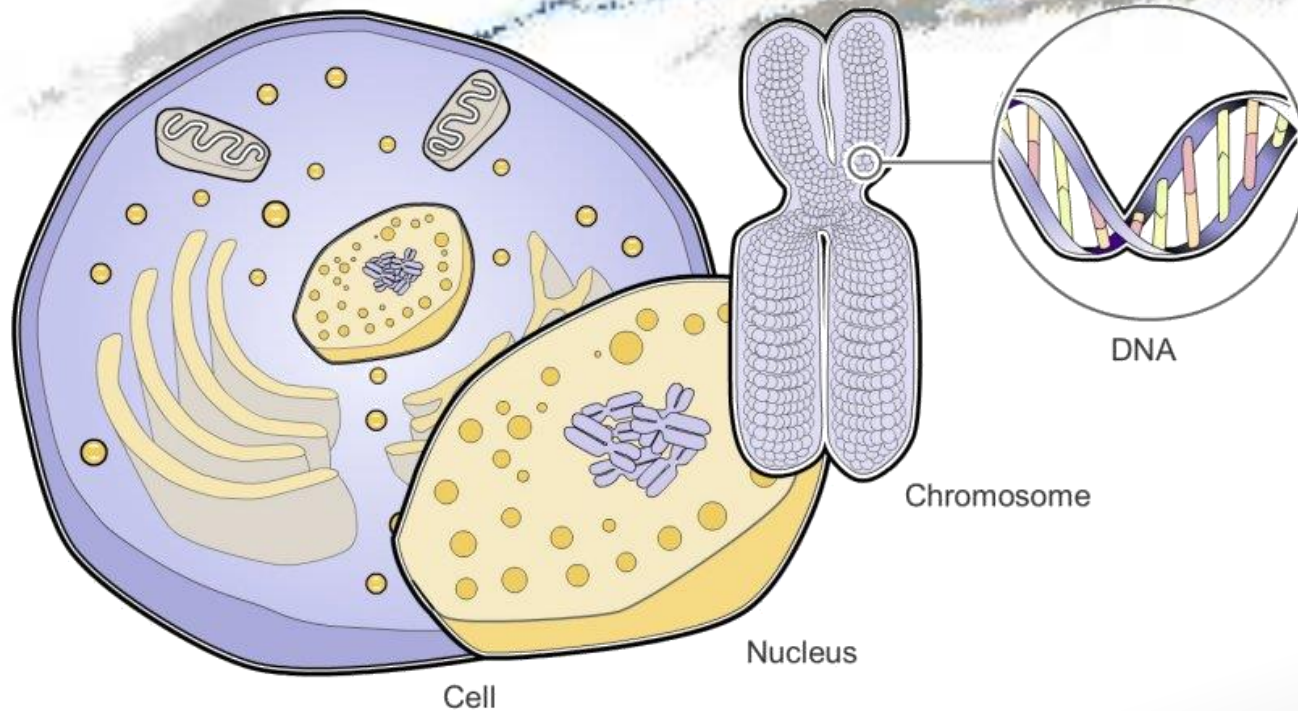


You must be familiar with the following genetic language and conventions:

gene, allele, mutation, genotype, phenotype, gamete, zygote, dominant, recessive, homozygous, heterozygous, pure breeding, Punnett square, and pedigree chart.

Genes are the sources of inherited information

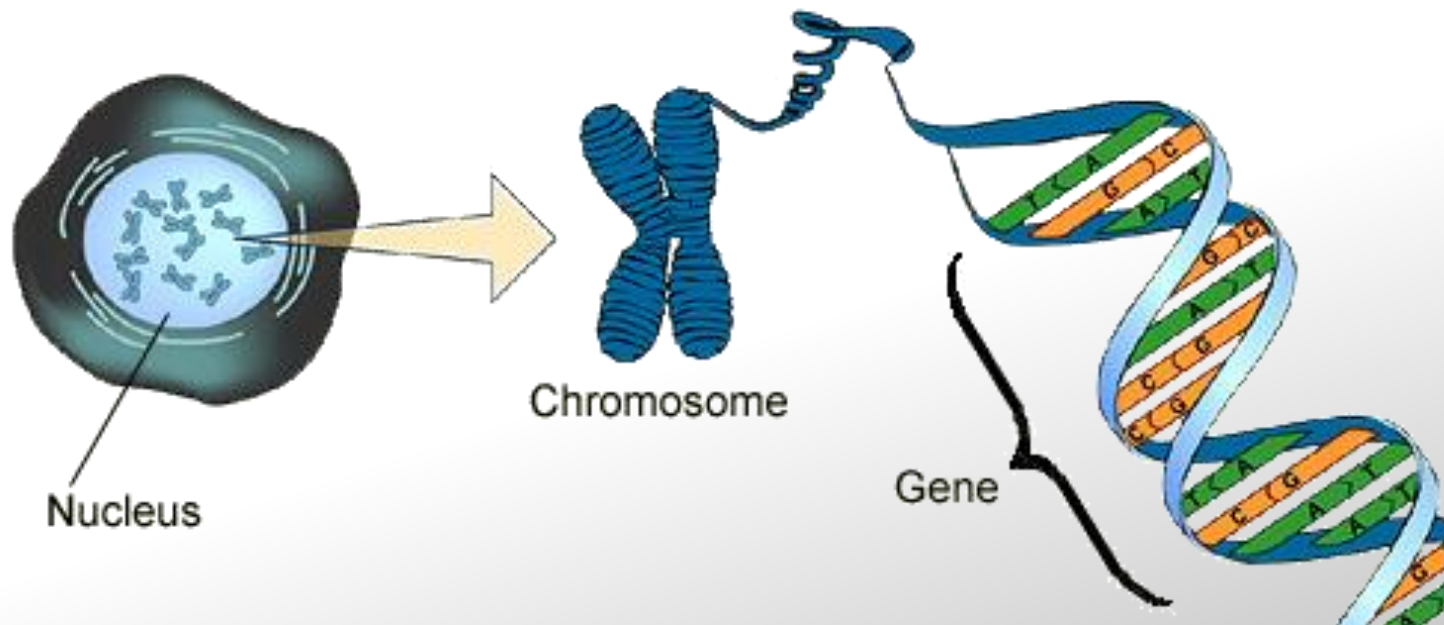
<https://www.yourgenome.org/video/from-dna-to-protein-flash>



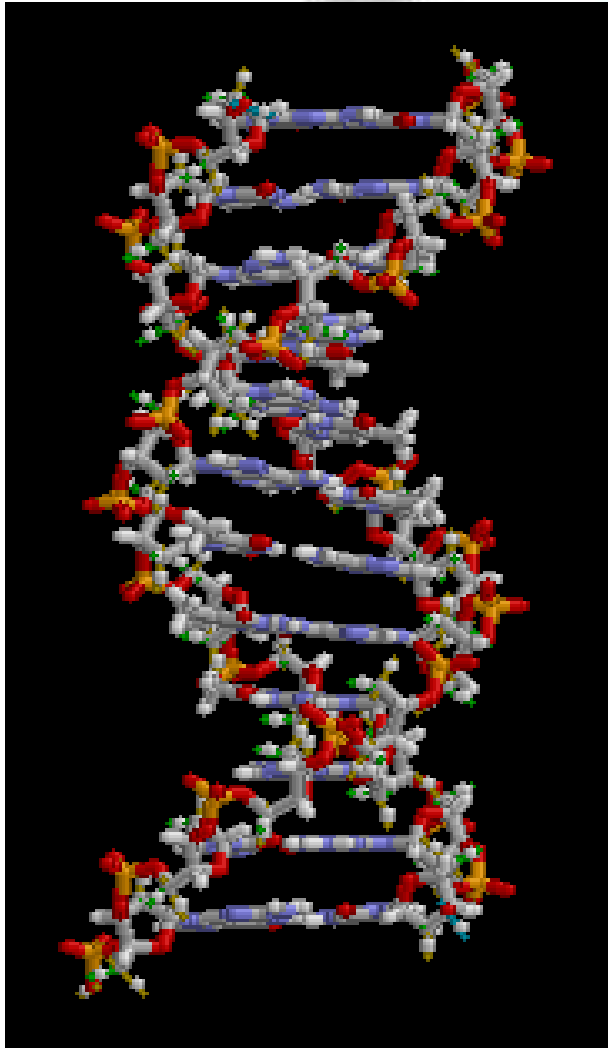
All living things are made of cells. The **nucleus** of a cell contains **chromosomes**, which carry instructions for the growth and development of an organism. The chromosomes are made of long strands of **DNA**. The order of molecules on the DNA strand code for protein. The instructions are called the **genetic code**. A segment of the DNA that codes for a specific protein is called a **gene**.

Chromosomes are found in the nucleus and genes are carried on chromosomes

DNA strands are loose within the nucleus of a cell. Just prior to cell division the DNA folds up into tight coils, then into structured chromosomes. The human cell has **46 chromosomes** arranged into 23 pairs of chromosomes. Each chromosome in a pair has the same genes, (called **homologous** pairs) – except the sex chromosome pair – although there may be variation between the genes of each pair, as one comes from the father and one comes from the mother.



Genes are 'coded instructions' for making proteins and that DNA is the chemical, which stores the coded instructions



DNA is arranged in a **double helix shape**. The up rights of the "ladder" consist of alternating **sugar** and **phosphate** molecules bonded together. Making up the "rungs" are two base molecules connected to each sugar molecule.

The **base** molecules are held together by hydrogen bonding which can be broken and then later reformed when the DNA molecule splits to make a copy for protein manufacture or DNA replication.



Genes are 'coded instructions' for making proteins and that DNA is the chemical, which stores the coded instructions

DNA (deoxyribonucleic acid) units are called **nucleotides** which consist of a sugar, a phosphate and a base.

There are 4 bases

A – Adenine

C – Cytosine

G – Guanine

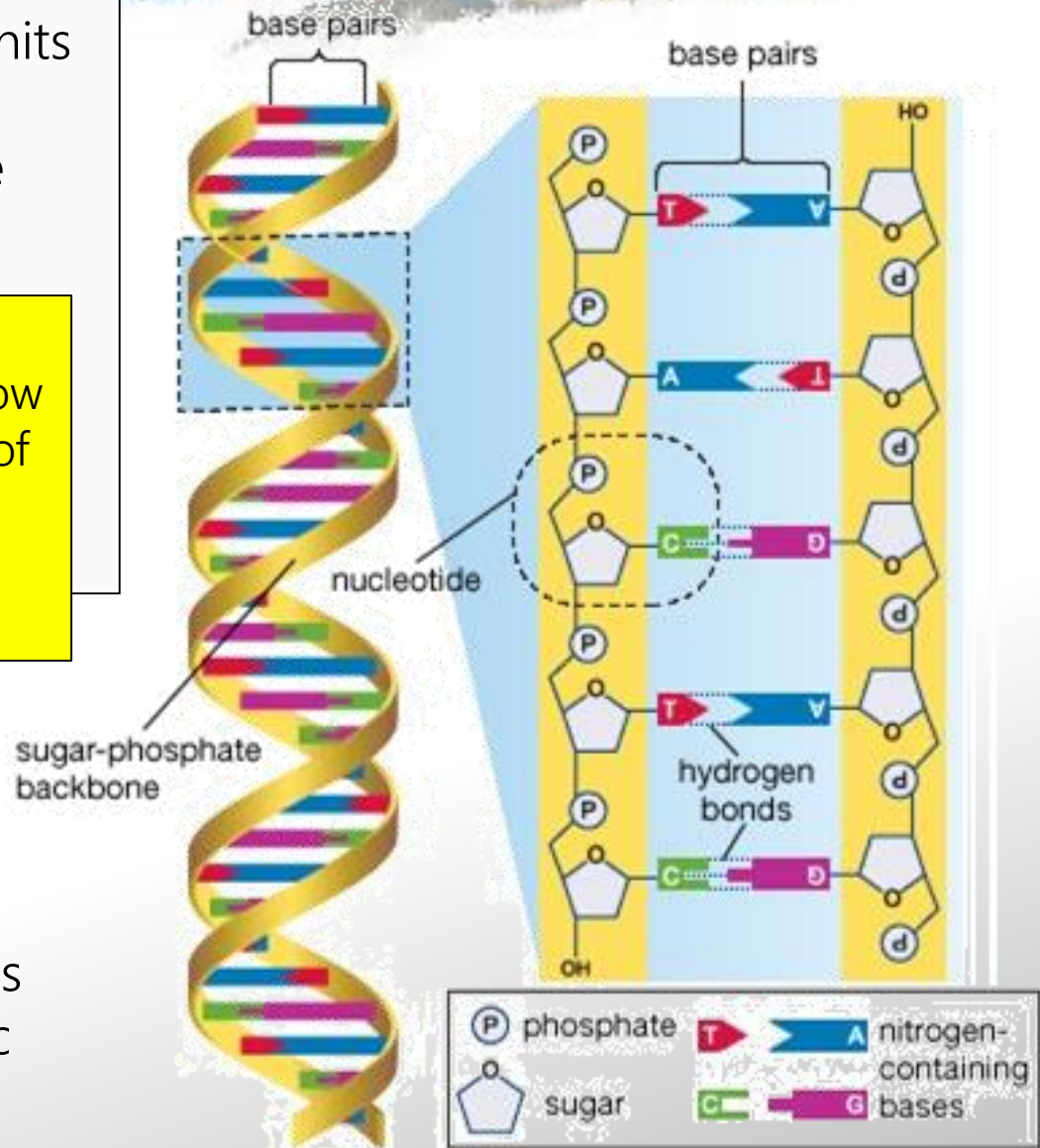
T – Thymine

You do not need to know the names of the bases – only their letters

G bonds with C

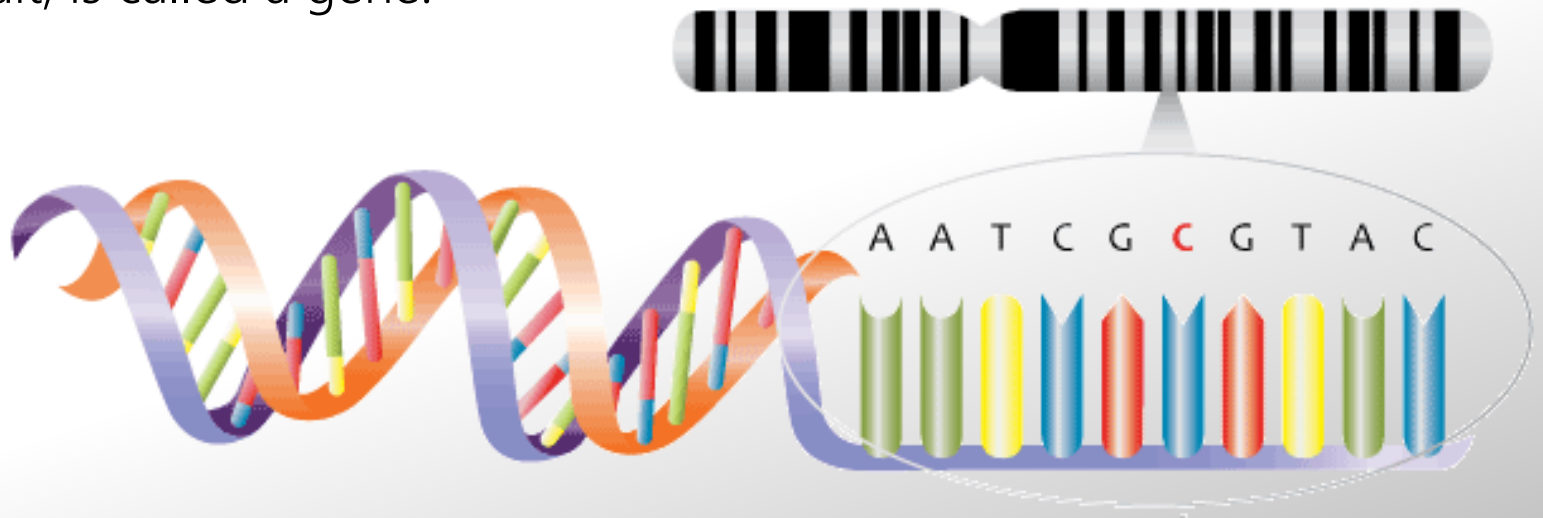
A bonds with T

This is called the **complimentary base-pairing rule**. The order of these bases is the **code** for producing specific proteins.



DNA Structure summary

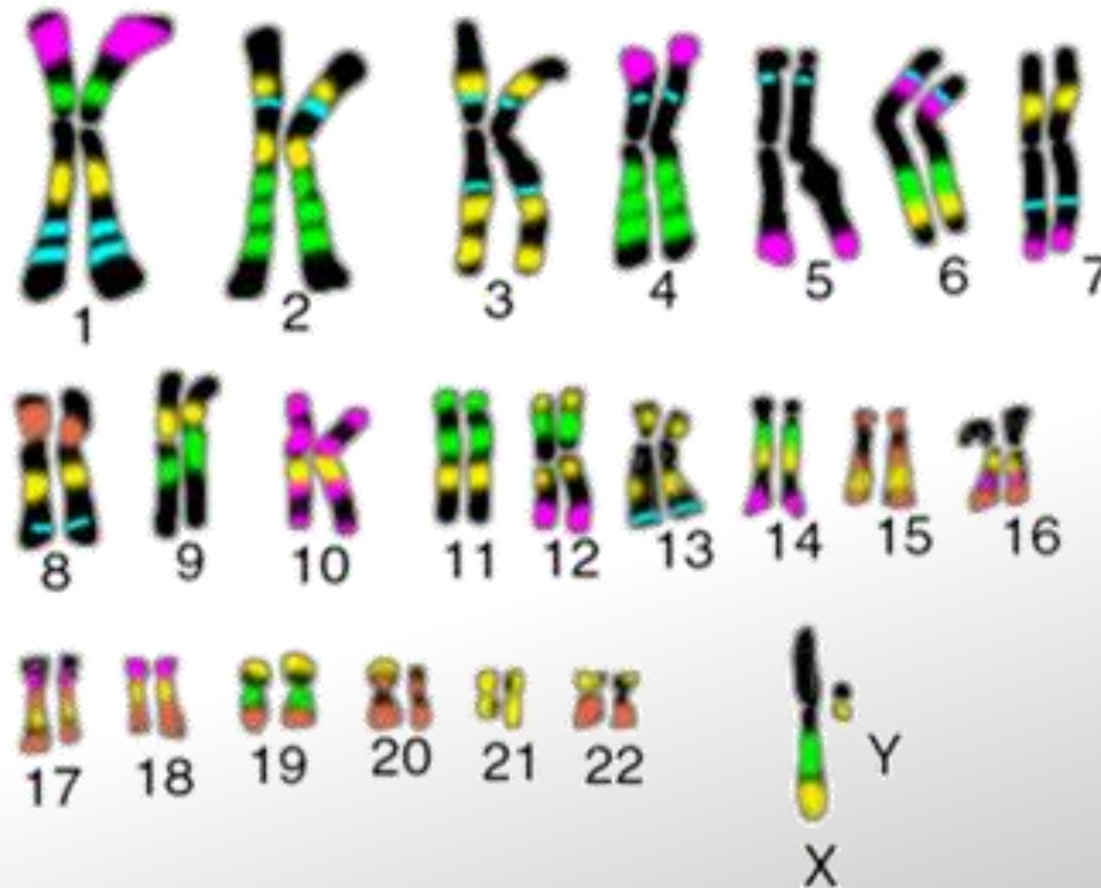
- ❑ DNA is made up of a series of nucleotides, each consisting of a sugar (deoxyribose) and base and a phosphate.
- ❑ DNA has four bases; A bonds to T and C bonds to G
- ❑ DNA is a double stranded helix that can separate during replication to produce 2 identical strands that allow DNA to be passed onto another cell or gamete.
- ❑ The base pairing rule makes DNA a stable molecule with less chance of mutation
- ❑ The sequence of bases coding for a specific proteins, leading onto a trait, is called a gene.



Human Karyotype contains a cells 'set' of chromosomes

A complete set of chromosomes of an organism placed into pairs of (**homologous**) chromosomes is called a **karyotype**. The human karyotype consists of 23 pairs of chromosomes. One pair is the sex chromosomes – XX in females and XY in males.

i
extra
info



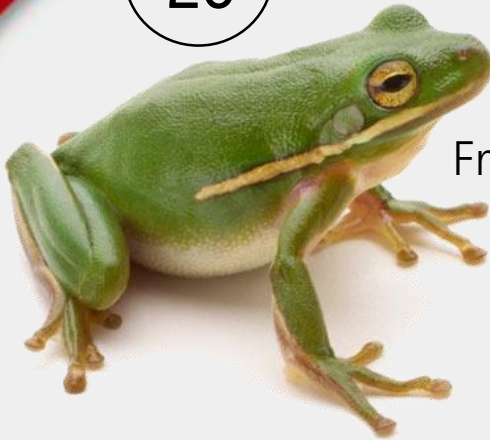
Background Knowledge

G2 Science Resources

Chromosome numbers of other species

Other species may have a different number of chromosomes in each cell compared to Humans. The number of chromosomes do not relate to the "complexity" of the organism.

26



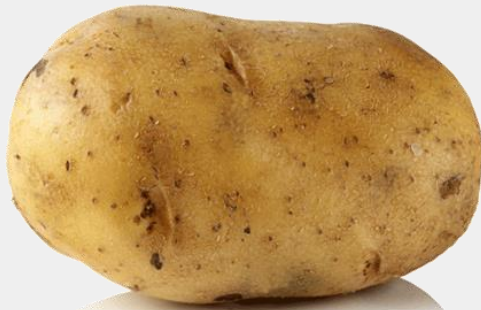
Frog

Pea

14



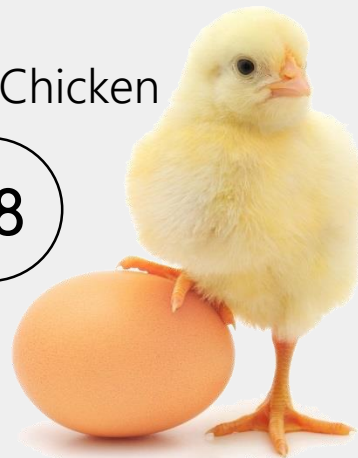
Potato



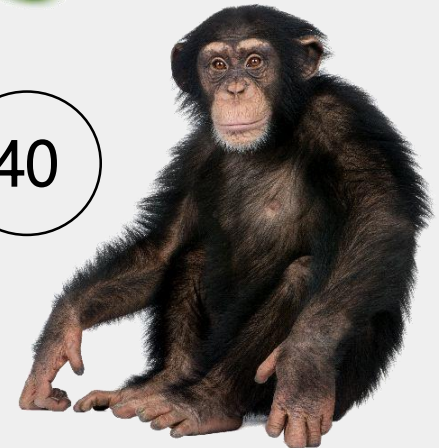
40

Chicken

78



40



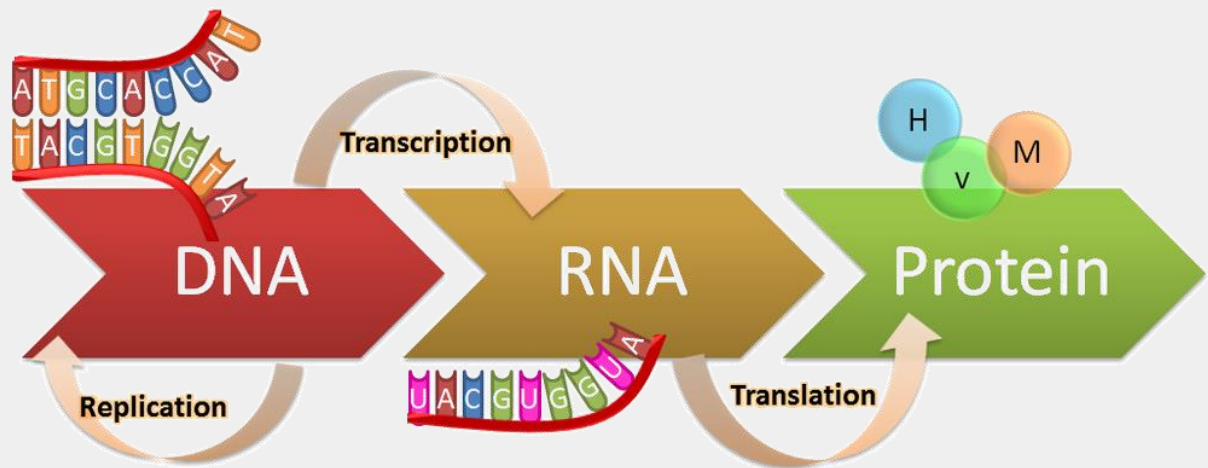
Chimpanzee



"Making protein"

The order that the bases are arranged in the DNA segment of a gene determine the type of protein that will be formed.

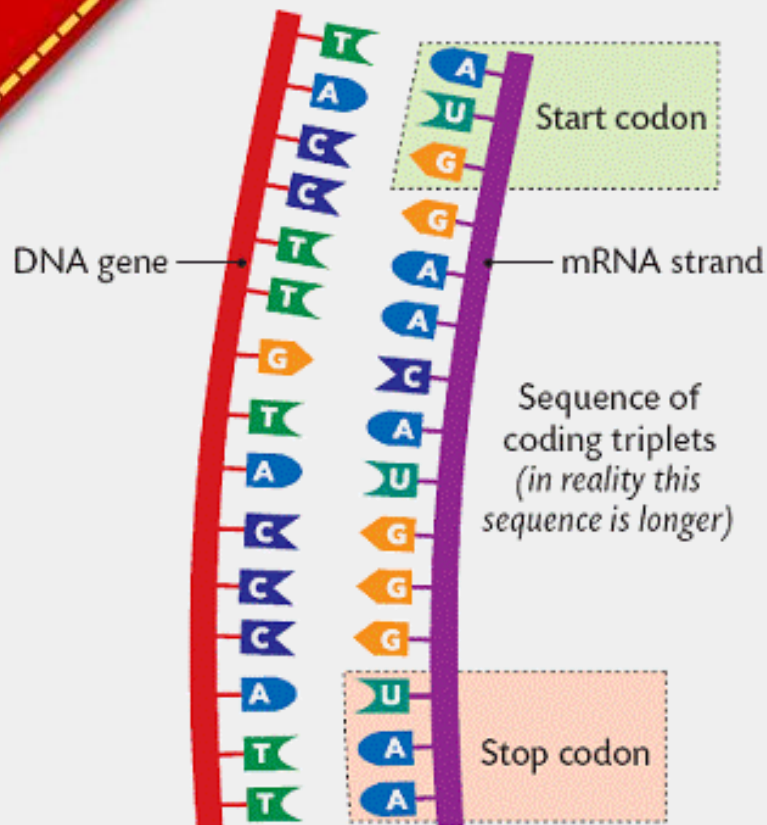
The bases are "read" in sets of three called **codons**. Codons are matched to molecules called amino acids which are the building blocks of proteins.



A **single stranded copy** of DNA from a gene is called **RNA**. (Transcription). The RNA moves out of the nucleus into ribosomes and the codons code for different amino acids which link together to form a protein molecule. (Translation)

Background Knowledge

Genes are 'coded instructions' for making proteins and that DNA is the chemical which stores the coded instructions

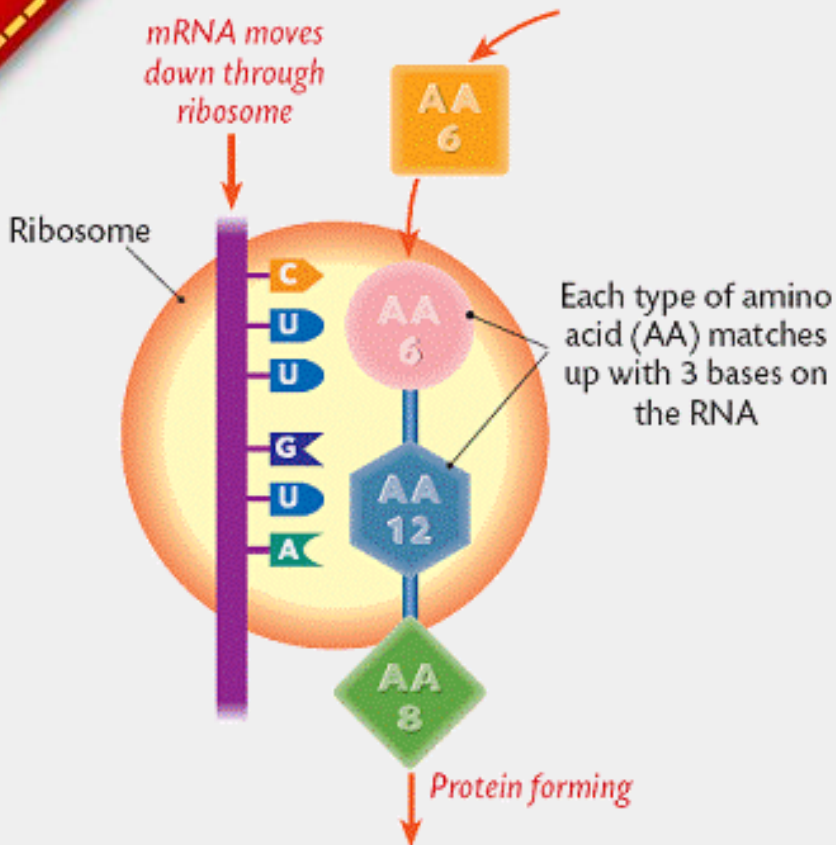


A **gene** is a code for one protein. When the cell requires a type of protein a copy of the particular gene is taken. **mRNA** (messenger RNA) is the name of the "photocopy" and it consists of a single strand of matching bases of the gene.

The mRNA then moves out of the nucleus and into a **ribosome** where the protein manufacture starts.

Background Knowledge

Genes are 'coded instructions' for making proteins and that DNA is the chemical which stores the coded instructions

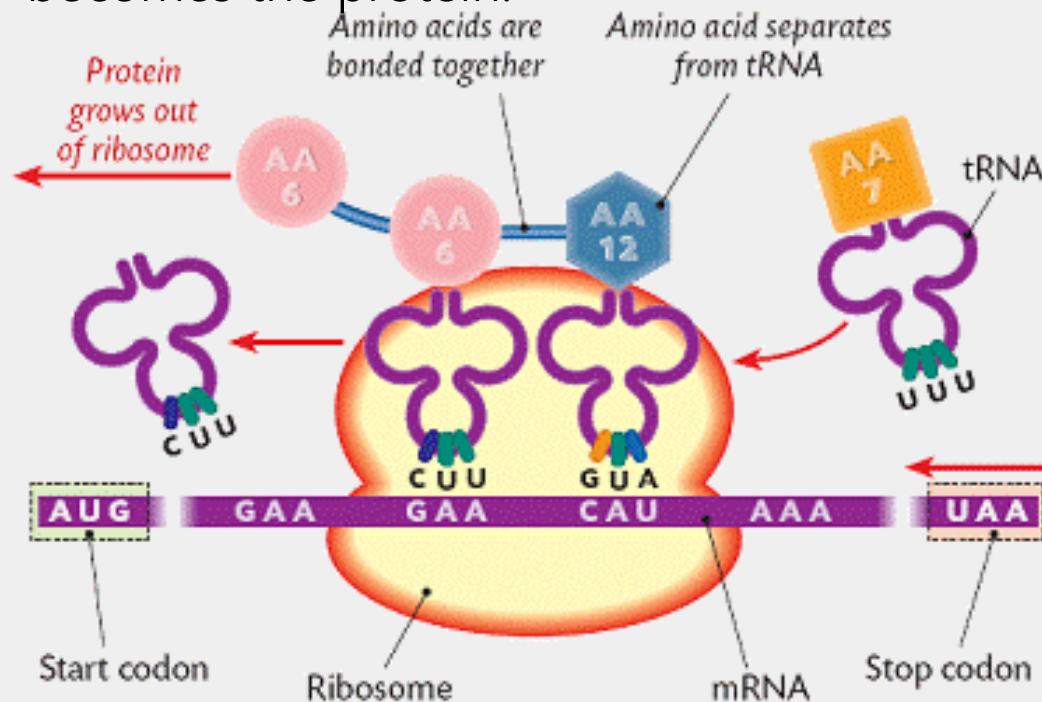


The mRNA moves through the ribosome as if it was on a conveyer belt. Each set of three bases called a **codon** codes for a particular type of amino acid to join to it. As each new amino acid is added it bond to the one beside it. This process continues until it reaches the end of the gene.

Background Knowledge

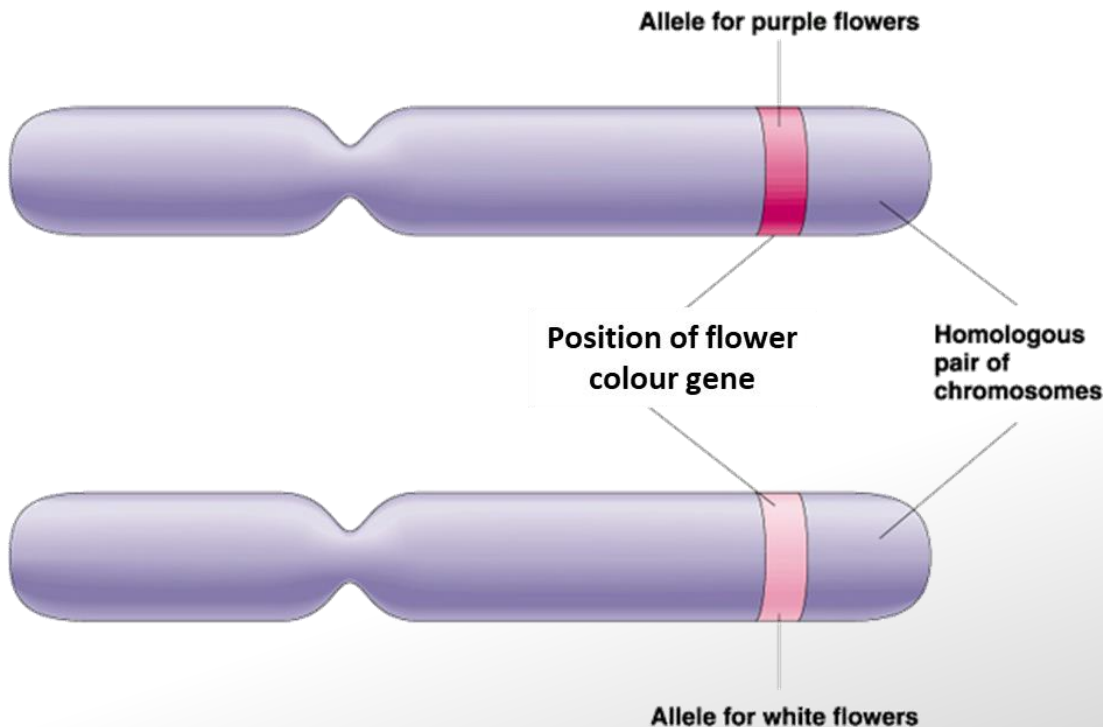
Genes are 'coded instructions' for making proteins and that DNA is the chemical which stores the coded instructions

Special molecules called **tRNA** (transport RNA) bring along the appropriate **amino acid**. There is one type of tRNA for each variety of codon. Once the chain is complete it is then folded up into particular shapes and becomes the protein.



Alleles are different versions of genes

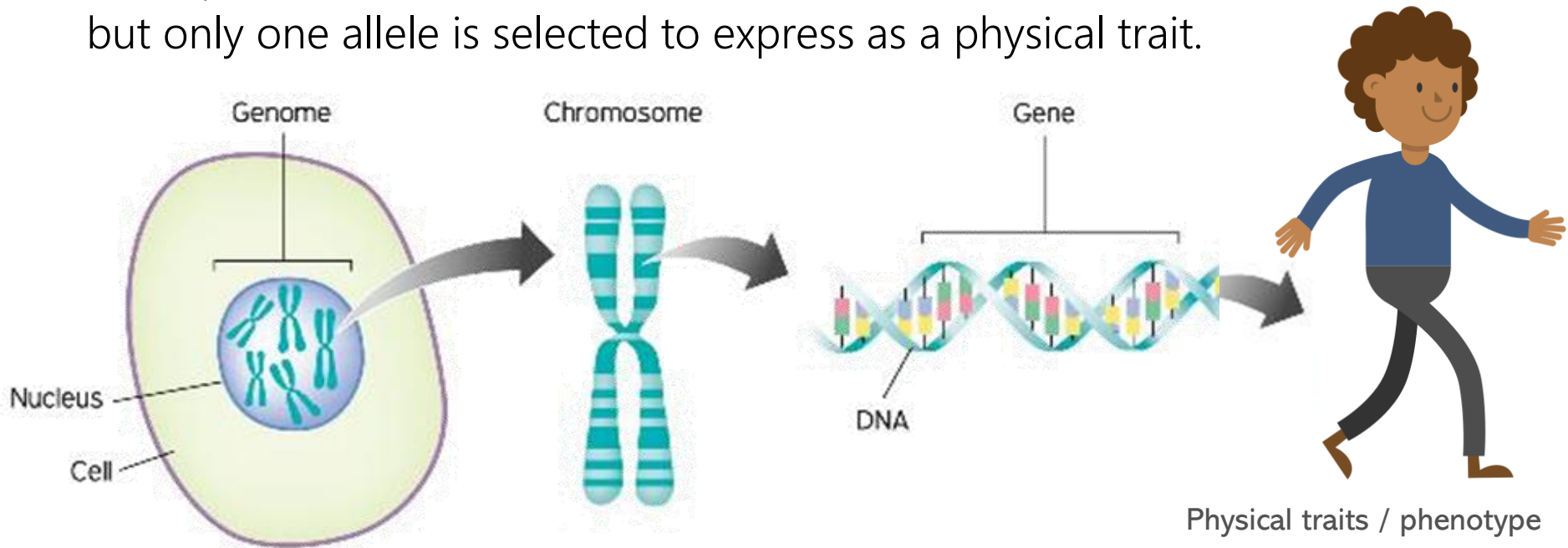
Chromosomes occur in (homologous) **pairs**. These pairs of chromosomes have genes that control the same traits at the same place (loci) in each. Genes may slightly differ from each other, due to base arrangement differences, and these versions of genes are called **alleles**. When the genes are being expressed only one allele needs to be used. The combination of alleles for any trait is known as the **genotype**. The physical trait expressed due to the genotype is known as the **phenotype**.



For example: The **gene** for flower colour is found in the same position on each of the chromosomes in a pair (one inherited from each parent). The different versions of genes, **alleles**, can create different physical traits, such as purple or white colour

Writing linking explanations between the nucleus, Chromosomes, DNA, genes and alleles

All living things are made of cells. The **nucleus** of a cell contains **chromosomes**, which carry instructions for the growth and development of an organism. The chromosomes are made of long strands of **DNA**. The order of molecules on the DNA strand code for traits/protein. The instructions are called the genetic code. A segment of the DNA that codes for a specific trait/protein is called a **gene**. Each gene is represented by two **alleles**, which are different varieties. The alleles can be the same or different but only one allele is selected to express as a physical trait.

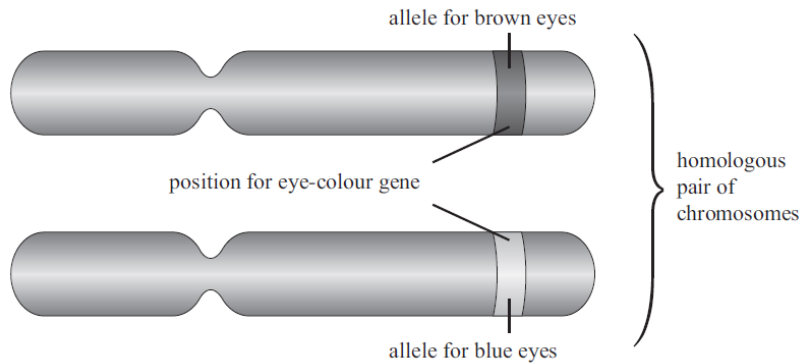


NCEA 2013 Genes and Alleles – Eye Colour

Excellence
Question

<http://www.nzqa.govt.nz>

Question 2a: Use the diagram below to help you explain the relationship between chromosomes, genes, alleles, phenotype, genotype, and the molecule DNA.



Answer 2a: Chromosomes are made up of DNA. DNA is a large molecule that is coiled into a double helix (twisted ladder structure). It is responsible for determining the phenotype of an organism. Along this molecule are bases. These bases pair up; A always pairs with T, and G with C. A sequence of bases which codes for a particular trait (eg, eye colour) is called a gene.

The different versions of each gene are called alleles, and these show the different variations of each characteristic, eg brown / blue eyes. Because chromosomes come in pairs for each trait, there will be two possible alleles. These different versions of genes (alleles) occur as the DNA base sequence is different.

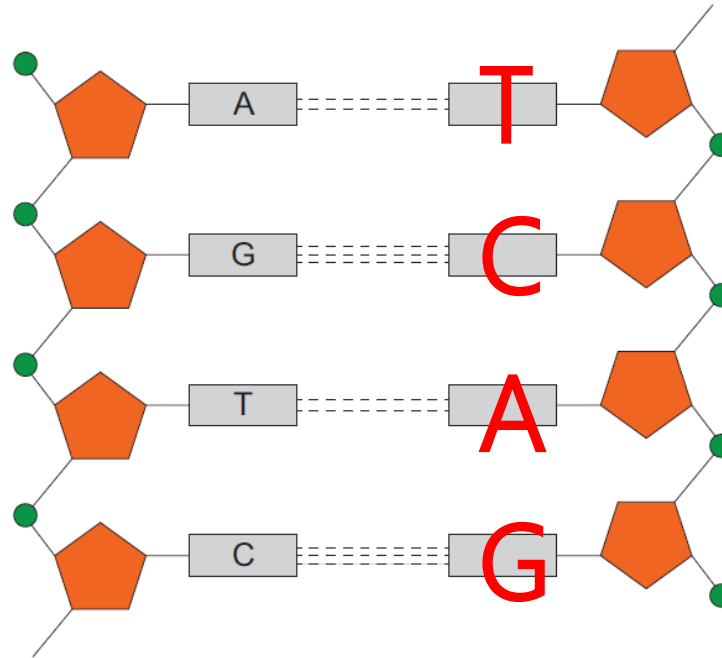
This combination of alleles for each trait is called the genotype; this can be any combination of two of the available alleles. The genotype determines the phenotype (the physical appearance) of the organism. Whichever alleles are present may be expressed. Dominant alleles (B) will be expressed over recessive alleles (b).

NCEA 2014 Genes and Alleles

<http://www.nzqa.govt.nz>

Achieved
Question

Question 1a: Label the unlabelled bases A, G, C, or T in the diagram of DNA shown below



Merit
Question

Question 1b: Explain the relationship between DNA, a gene, and an allele.

Answer 1b: A gene and an allele are both made of DNA. A gene is a length of DNA that codes for a particular characteristic, whereas an allele is the alternative form of a gene containing a different base sequence. A gene for a characteristic consists of two alleles. The dominant allele gives the phenotype of the gene.

NCEA 2015 Genes and Alleles – Snails (part ONE)

<http://www.nzqa.govt.nz>

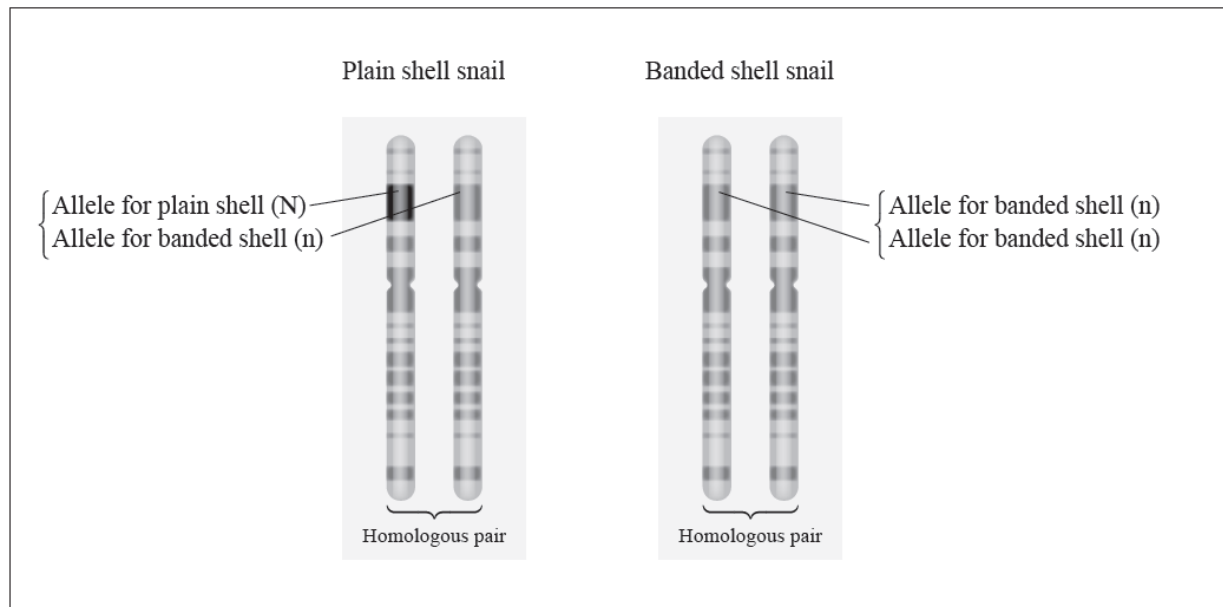
Excellence
Question

Question 2a: A snail known as *Cepaea nemoralis* can have either a plain shell or a banded shell.



The diagrams below show the homologous chromosomes that contain the gene for shell pattern for each of the snails in the photographs above.

Assume the allele for plain shell (N) is dominant over the allele for banded shell (n).



NCEA 2015 Genes and Alleles – Snails

<http://www.nzqa.govt.nz>

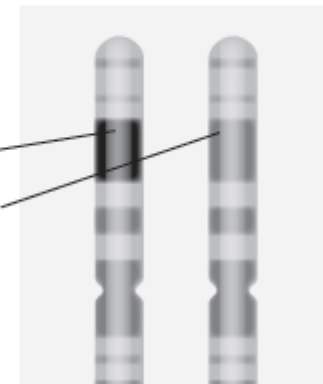
Achieved
Question

Question 2b: In the diagram on the previous page, which snail is heterozygous for shell pattern?

Explain why you chose this snail.

Answer 2b: Plain shell is heterozygous for shell pattern.
The alleles are different from each other

Plain shell snail



{ Allele for plain shell (N)
{ Allele for banded shell (n)



Merit
Question

Question 2c: Referring to the examples shown previously for shell pattern, explain the difference between an allele and a gene.

Answer 2b: A **gene** is a length of DNA that codes for a particular characteristic protein such as shell pattern, whereas an **allele** is an **alternative form of a gene**. There is one allele for a banded shell and a different allele for a plain shell. The two alleles together make up the genotype.

NCEA 2016 Genes and Alleles – Rock pocket Mice

Excellence
Question

<http://www.nzqa.govt.nz>

Question 2a: Rock pocket mice can have dark fur or light fur, as shown below. Using the example of rock pocket mouse fur colour, explain how information carried on the DNA controls the appearance.

In your answer you should refer to DNA base sequence, genes and alleles.

Answer 2a: DNA is the (molecule) that carries the genetic code/ **information**. The base sequence is the order of bases (A, T, C and G) that carry the code. A **gene** is a section of DNA that codes for 1 trait /_protein, e.g. mouse fur colour. An **allele** is a gene form, e.g. light or dark fur.

A difference in the DNA results in a difference in how the genetic information is read and can create a different appearance (phenotype). This is a different gene form (allele). In this case, the light coloured mouse would have a different base sequence to the dark mouse on the gene for fur colour.



Light colour

<https://inaturalist.org>



Dark colour

Question 3b: Explain how the sexual reproduction of kauri trees causes genetic variation AND how this could lead to increased survival of the species when faced with kauri dieback disease.

In your answer you should consider:

- the processes of gamete formation (meiosis) and fertilisation
- how sexual reproduction leads to variation in the population
- the link between genetic variation and the survival of kauri trees as a species.

<http://www.nzqa.govt.nz>

Gamete formation by meiosis: Random assortment/segregation / crossing of chromosomes over in meiosis

Description of meiosis: produces gametes / sex cells that have half the normal number of chromosomes as body cells.

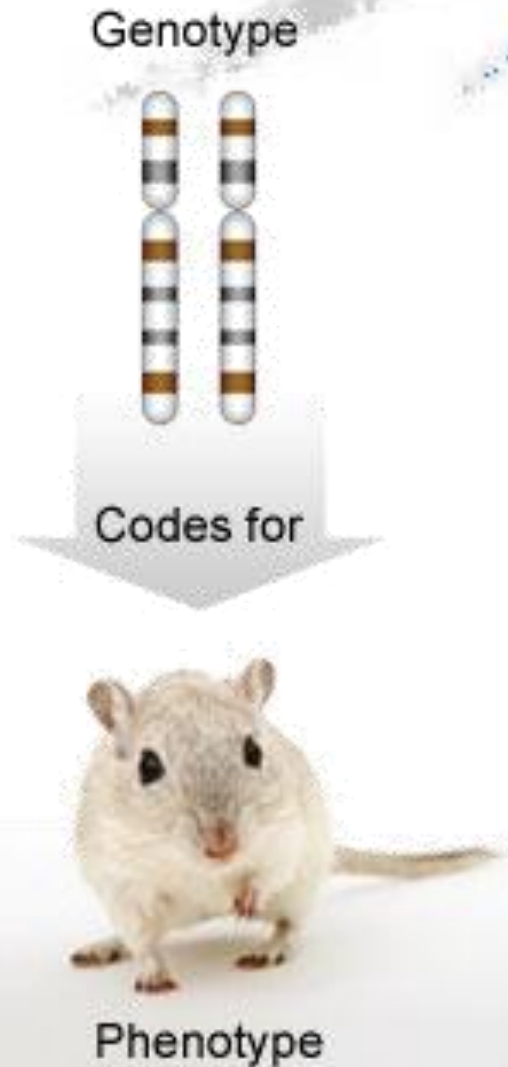
Process of fertilisation: Random male and female gametes join, each with unique DNA producing a genetically unique zygote / offspring.

Role of sexual reproduction: produce new combinations of alleles and thus genetic variation between individuals.

Explanation: The advantage of genetic variation to a species is that it may enable some individuals to survive kauri dieback to reproduce, passing on favourable alleles / genes to the next generation. Over many generations this genetic advantage / genes / alleles will rise in the population, allowing survival of the kauri species.





Phenotype and genotype

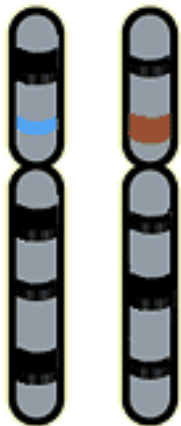


The **genotype** is the combination of 2 alleles that an organism contains for a trait on its two homologous chromosomes. For any particular trait they can be heterozygous (different) or homozygous (same). The **phenotype** is the physical trait that results because of the particular combination of alleles (genotype)

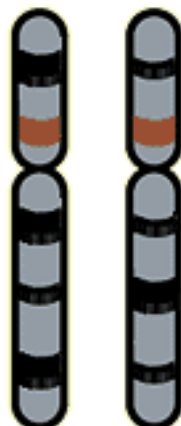
Dominant and recessive alleles

The allele that is expressed as a trait when there is at least one allele is called the **dominant** allele. The allele that is expressed if a dominant allele is not present is called the **recessive** allele. When there are two of the same alleles, this genotype is called **homozygous**. When there are two different alleles in a genotype, this is called **heterozygous**. The dominant allele is always expressed.

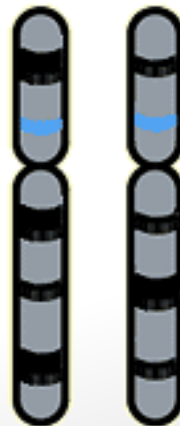
 = allele for blue eyes (recessive)
 = allele for brown eyes (dominant)



Individual A:
heterozygous



Individual B:
homozygous



Individual C:
homozygous
recessive

Phenotype = Blue Eyes



Genotype = bb
Recessive = b

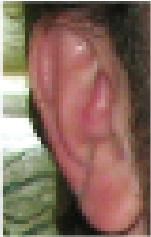
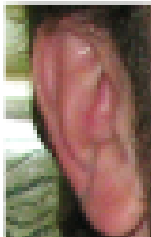
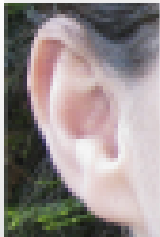
Phenotype = Brown Eyes



Genotype = Bb or BB
Dominant = B

Recording Genotype and Phenotype

A pair of letters is used to indicate the genotype – there are three possible genotypes if a dominant and recessive allele exist. Phenotypes are recorded as physical descriptions – there are two possible phenotypes, either the dominant or recessive

Genotype	Phenotype
EE Homozygous dominant	Detached Earlobes 
Ee Heterozygous	Detached Earlobes 
ee Homozygous recessive	Attached Earlobes 

Many letters can be used to represent dominant or recessive, such as Bb, Ee, Ff as long as the upper and lower case letters look different.

The genotype determines the phenotype.

Lop eared rabbits – an example

Rabbit ears normally point straight up. Some rabbits have an allele for lop ears that cause the ears fold down. The allele that produces lop ears is recessive and was created by a mutation.



lop eared rabbit



straight eared rabbit

We can use the symbol R to show the dominant allele and r to show the recessive allele.

The genotype of the two rabbits if both are **pure breeding** are:

Lop eared rabbit genotype
rr

Normal eared rabbit
genotype RR

Putting it all together – Genetics Vocabulary

Chromosomes are made up of **DNA**. DNA is a large molecule that is coiled into a double helix (twisted ladder structure). It is responsible for determining the **phenotype** of an organism. Along this molecule are **bases**. These bases pair up; A always pairs with T, and G with C.

A sequence of bases which codes for a particular **trait** (e.g., eye colour), is called a **gene**. Genetic information is determined by the order of the bases.

The different versions of each gene are called **alleles**, and these show the different **variations** of each characteristic, e.g. brown / blue eyes. Because chromosomes come in pairs for each trait, there will be two possible alleles. These different versions of genes (alleles), occur as the DNA base sequence is different.

This combination of alleles for each trait is called the **genotype**; this can be any combination of two of the available alleles. The genotype determines the phenotype (the physical appearance) of the organism. Whichever alleles are present may be expressed. **Dominant** alleles (B) will be expressed over **recessive** alleles (b).

NCEA 2013 Genotype and Phenotype – Eyes

Excellence
Question

Question 2b: The allele for brown eyes (B) is dominant over the allele for blue eyes (b) in humans.

Discuss how it would be possible for a child to have blue eyes, even though both their parents have brown eyes. In your answer you should:

- use labelled Punnett squares
- link the genotypes and phenotypes of the child, parents, AND grandparents.

Answer 2b: For the child to have blue eyes they must have a genotype of bb (i.e. have both recessive alleles). If a dominant allele, B is present then brown eyes would be seen.

In order to have a genotype of bb, each parent must have given a b (recessive allele). Both parents have brown eyes so therefore they both must have a dominant allele (B) and because each parent passes on a recessive allele the genotype of each parent must be Bb. The grandparents could have a genotype of bb, Bb, or BB. It is not possible to say for sure, but at least one of the grandparents on each side must pass on a recessive allele (b) in order for each parent to have a recessive allele to pass on to the child. Punnett square(s) may be used to show this but must be explained.



NCEA 2014 Genotype and Phenotype – Brothers

Achieved
Question

Question 2d (ii) : Two brothers, who have the same parents and are not identical twins, will have different genotypes and phenotypes. Define the term genotype. Define the term phenotype.

Answer 2d (ii) : Genotype – The combination of alleles for each trait.

Phenotype – The physical expression of genotype, alleles (the characteristic that is seen), e.g. blue eyes or brown eyes.

Question 2d(iii): Explain how the two brothers with the same parents can have different genotypes.

In your answer you should explain:

- the importance of meiosis
- the role of fertilisation.

Excellence
Question

Answer 2d(iii): The brothers will have **inherited half their chromosomes** from their mother and half from their father. The actual chromosomes they inherited are due to chance because **meiosis** (production of gametes) randomly mixes chromosomes, resulting in a new mix of genetic information, and therefore different genotypes and phenotypes. **Fertilisation** (fusion of parents gametes) is also random, with more variation occurring, depending on which sperm fertilises which egg, and because each sperm and egg are different in terms of which chromosomes are present, more variation is introduced in terms of each zygote having different combinations of chromosomes.

NCEA 2015 Genotype and Phenotype – Snails

Question 2c: These two snails were produced by sexual reproduction from the same male and female.

Discuss how they have inherited different alleles for shell pattern.

In your answer you should:

- explain where the homologous chromosomes have come from
- give the possible genotypes of both parents and explain how you determined these possible genotypes.

Answer 2c: The snails have inherited different shell patterns because they have inherited one homologous chromosome from their mother and one from their father. The banded snail is homozygous for shell pattern; therefore each parent must have contributed a banded allele. The plain snail has one plain allele; therefore at least one of the parents must have a plain allele. This means there are two possibilities for the parents: they are either both heterozygous; or one parent is heterozygous and the other is homozygous recessive.

In a homologous pair, one chromosome has come from the mother and one has come from the father. Because the banded snail has two banded alleles, each parent must have a banded allele.

Because the plain snail is heterozygous, at least one of the parents must have an allele for a plain shell.

There are two possibilities: the parents could both be heterozygous; or one parent could be heterozygous and the other could be homozygous recessive

NCEA 2016 Genotype and Phenotype – Rock pocket Mice

Question 2b: In rock pocket mice, dark fur colour (D) is dominant to light fur colour (d). Each mouse has two alleles for fur colour.

Explain how they inherit these two alleles, and explain how the two alleles interact to produce different phenotypes. In your answer you should:

- define phenotype and genotype
- explain how the alleles are inherited from the parents
- state the three possible fur colour genotypes for rock pocket mice.

Answer 2b: Phenotype is the appearance of a trait, e.g. dark or light fur. Genotype is the code for the alleles present for the gene. Each mouse receives one copy of each gene from each of its parents during fertilisation, one from the sperm and one from the egg, and so has two copies of each. If either of these are the dominant allele (i.e. DD or Dd), the mouse will be dark, as the dark allele is dominant (and so masks the light allele). If both are recessive (dd), the mouse will be light.

link the inheritance of one copy of each allele from each parent combines (through the sperm and egg / fertilisation) AND how these alleles interact via three genotypes to give the two phenotypes, with reference to dominant alleles



Gene pool

The gene pool is all of the present alleles (versions of genes) present in a population. Theoretically all alleles present are then available to be passed on to further generations through breeding. Generally the larger a population the larger (or more diverse) a gene pool will be.

*A **gene pool** is the sum of all the individual genes in a given population.*





Genotype Frequency

The frequency that a genotype (AA – homozygous dominant, aa – homozygous recessive, Aa – heterozygous) is present in a population for a given gene is known as its genotype frequency.



The frequency of a genotype is the proportion that is present in a population compared to all other possibilities.

Background Knowledge

Genotype Frequency

Population total = 10

Homozygous dominant (AA) = $2/10 \times 100 = 20\%$

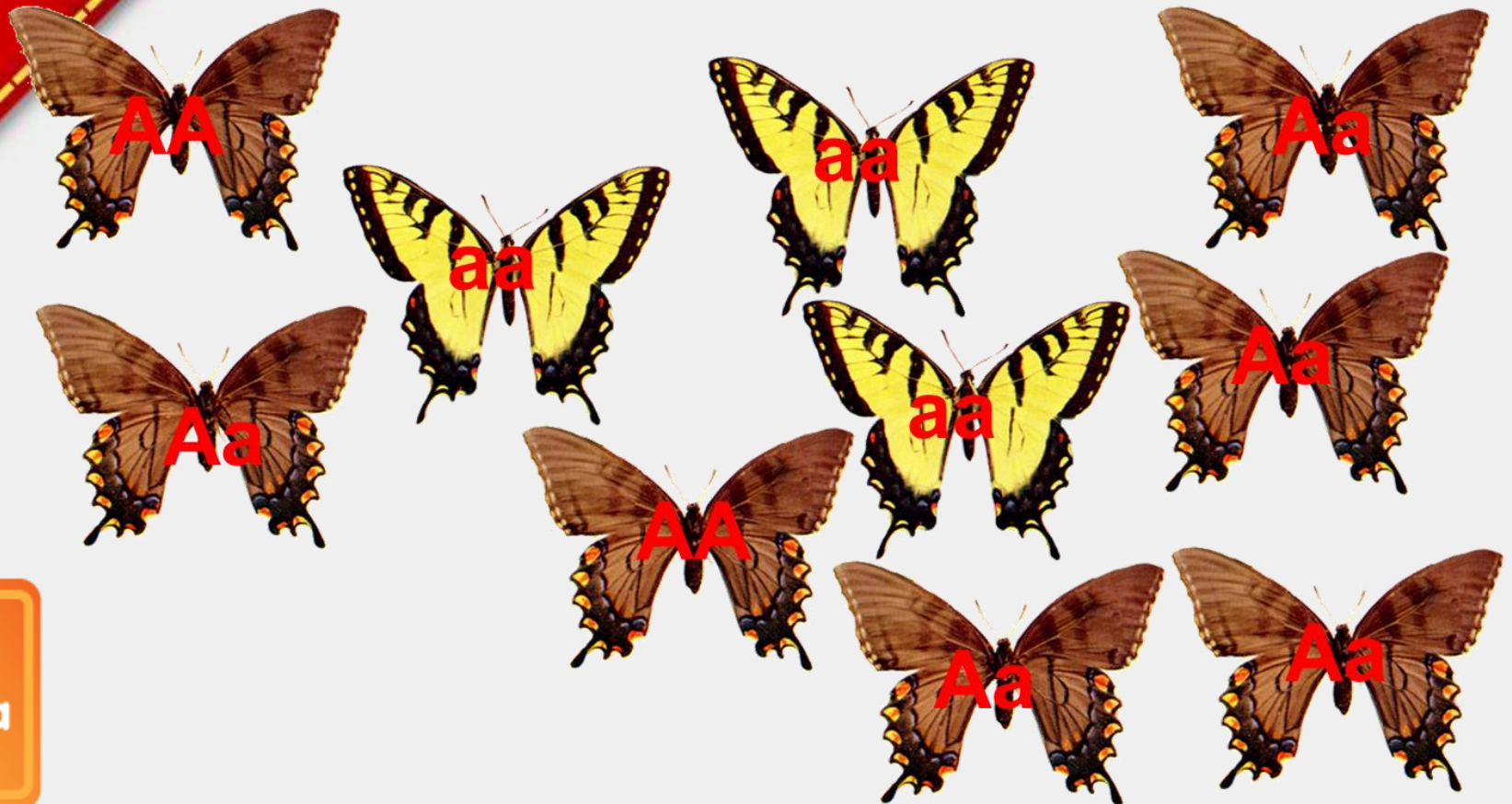
Homozygous recessive (aa) = $3/10 \times 100 = 30\%$

Heterozygous (Aa) = $5/10 \times 100 = 50\%$



Phenotype Frequency

The phenotype is the trait that we can see, in this case brown colour (dominant A) or yellow colour (recessive a). Phenotype for yellow = $\frac{3}{10} \times 100 = 30\%$, phenotype for brown = $\frac{7}{10} \times 100 = 70\%$

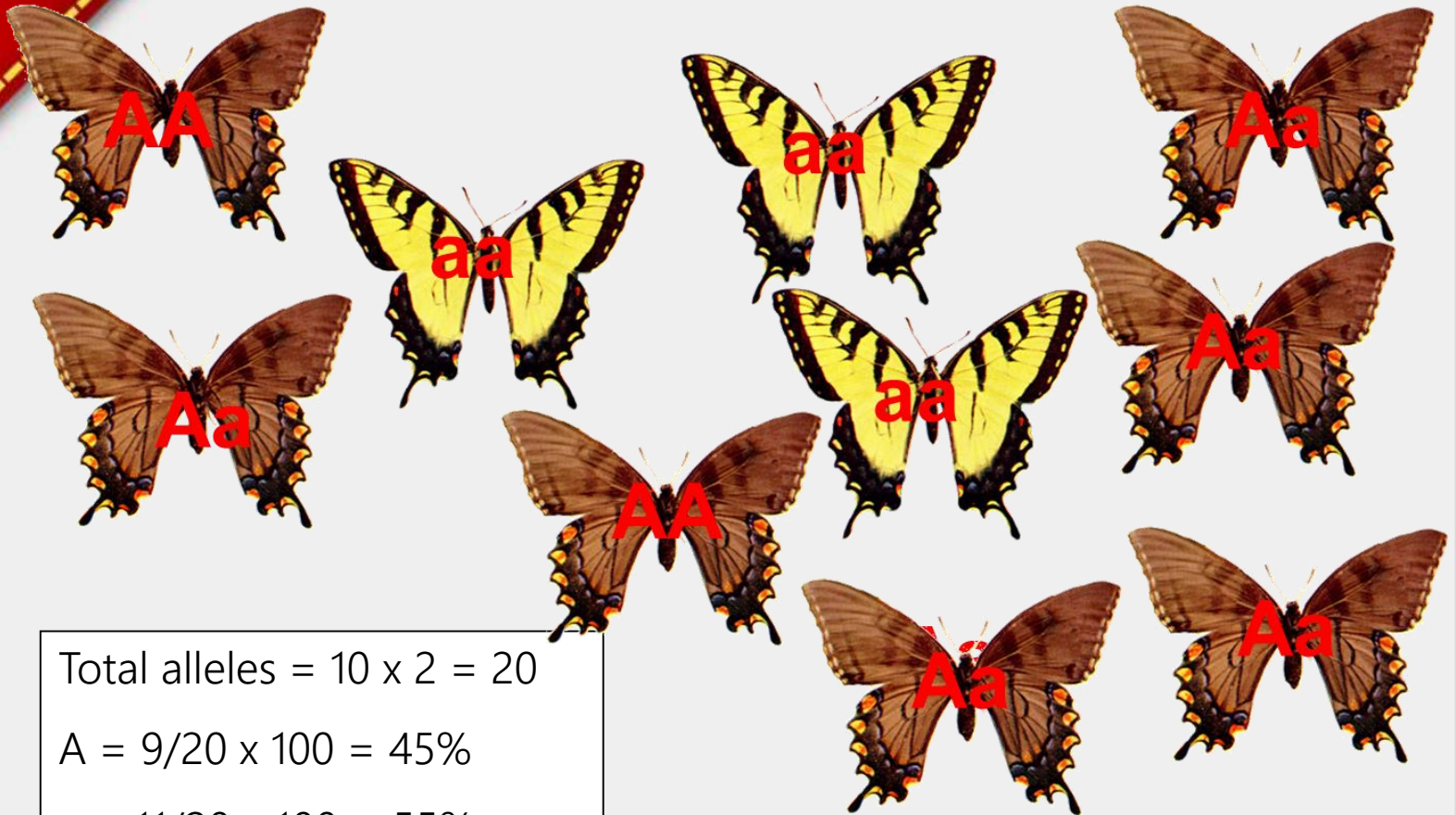


Background Knowledge



Allele Frequency

The frequency that an allele is present in a population compared to all other alleles of the same gene, is known as the allele frequency.



$$\text{Total alleles} = 10 \times 2 = 20$$

$$A = 9/20 \times 100 = 45\%$$

$$a = 11/20 \times 100 = 55\%$$



extra
info

Gregor Mendel

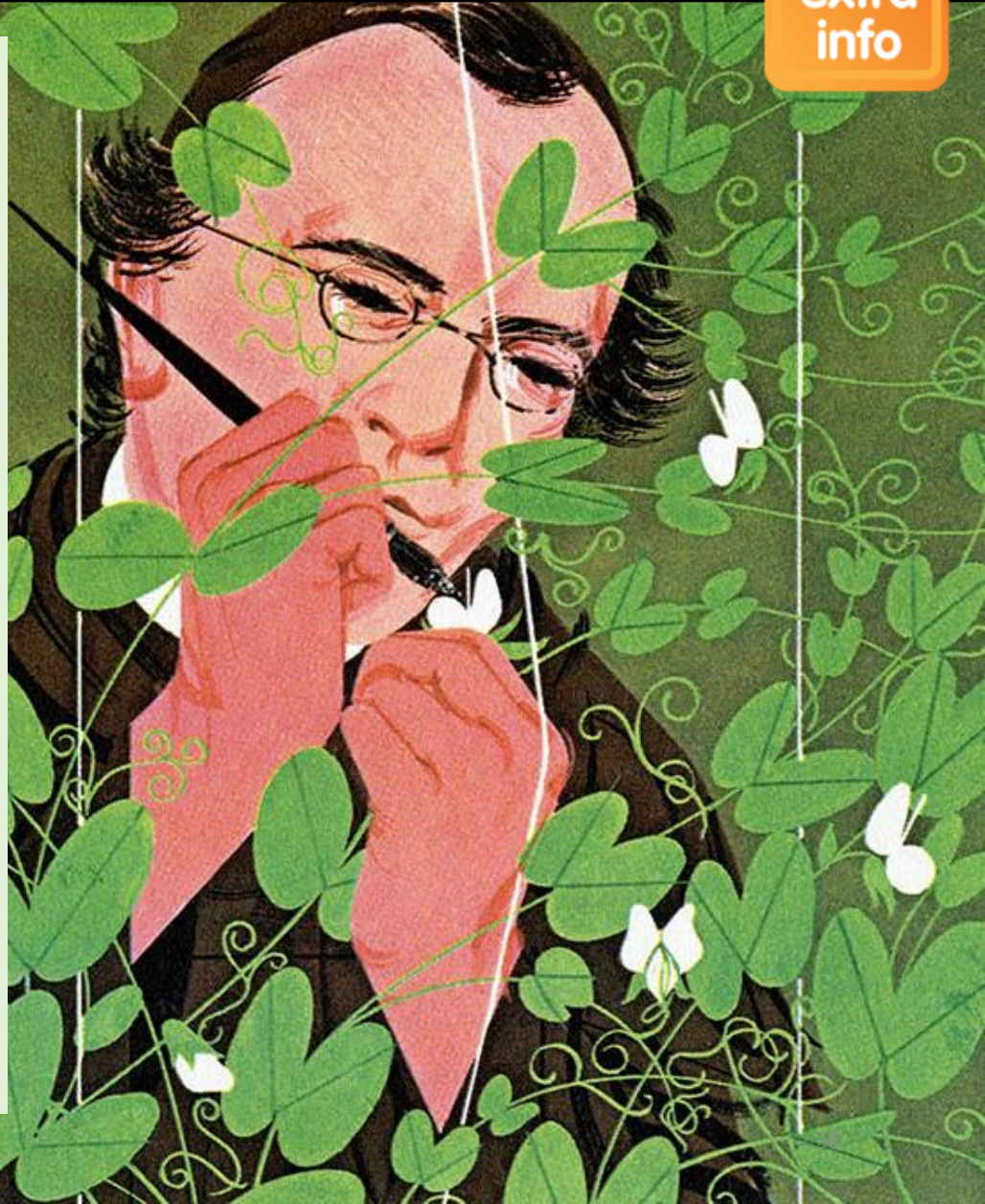


Gregor Mendel was a German Friar who lived from 1822-1884. Mendel is called the “Father of Genetics” because through his work on over 10,000 pea plants he discovered the main laws of inheritance.

1) **The Law of Segregation:** Each inherited trait is created by a gene pair. Adult genes are randomly separated to the gametes and gametes contain only one gene of the pair. Offspring inherit one genetic allele from each parent when gametes combine in fertilisation.

2) **The Law of Independent Assortment:** Genes for different traits are sorted separately from one another

3) **The Law of Dominance:** An organism with different alleles of a gene will use the allele that is dominant.



Using Punnett squares to predict offspring

We use Punnett squares to keep track of alleles when calculating the genotype of any offspring created when two organisms are mated



Agouti
Rabbit

B

b

b

Bb

bb

b

Bb

bb



Black Rabbit

B is the
dominate allele
for Agouti
colour.

b is the
recessive allele
for Black colour

Each adult gives
one allele to
each offspring.

How to use a Punnett squares to predict offspring



wiki How to Make a Punnett Square

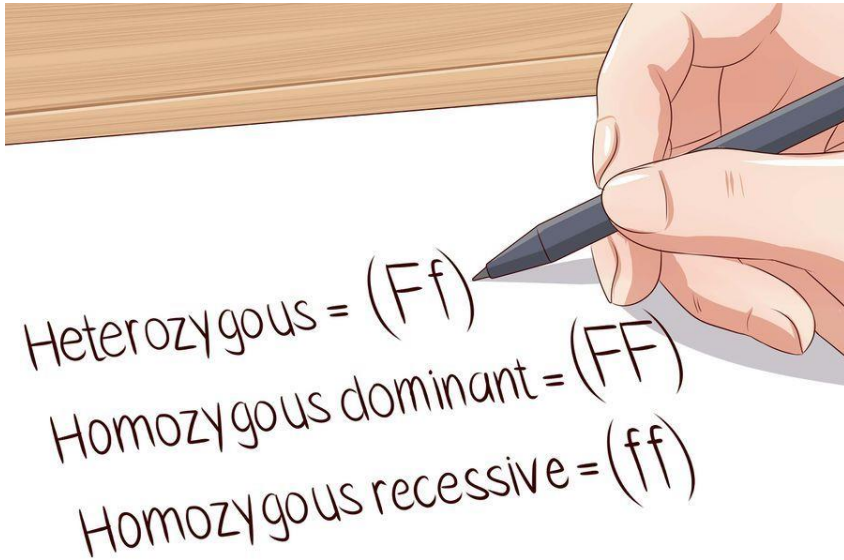
1. Draw a grid with 4 squares. Each square will represent 1 out of 4 offspring or 25% of offspring out of 100%

2. Write down the possible 2 phenotypes (physical traits) and label them with a capital letter for the dominant trait and lower case letter for the recessive trait.



wiki How to Make a Punnett Square

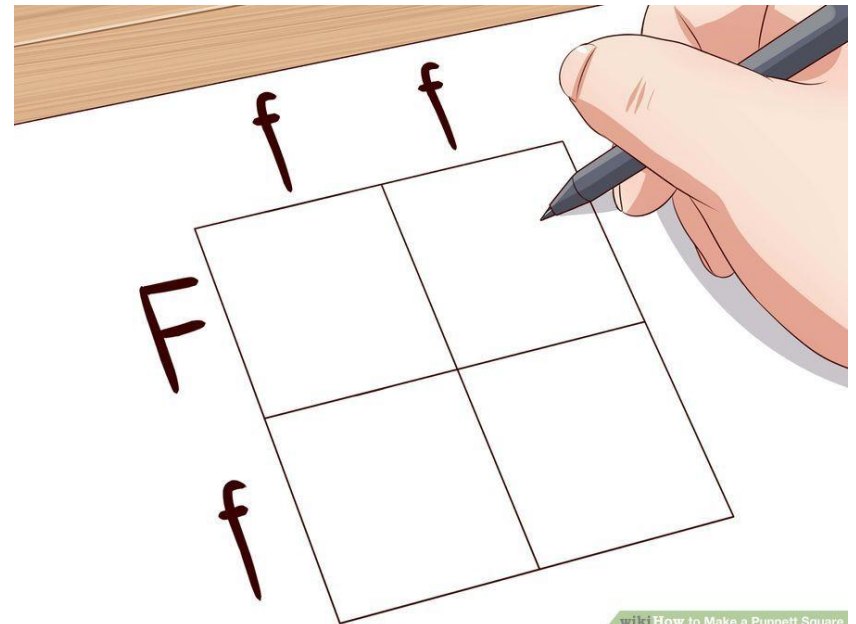
How to use a Punnett squares to predict offspring



wikiHow to Make a Punnett Square

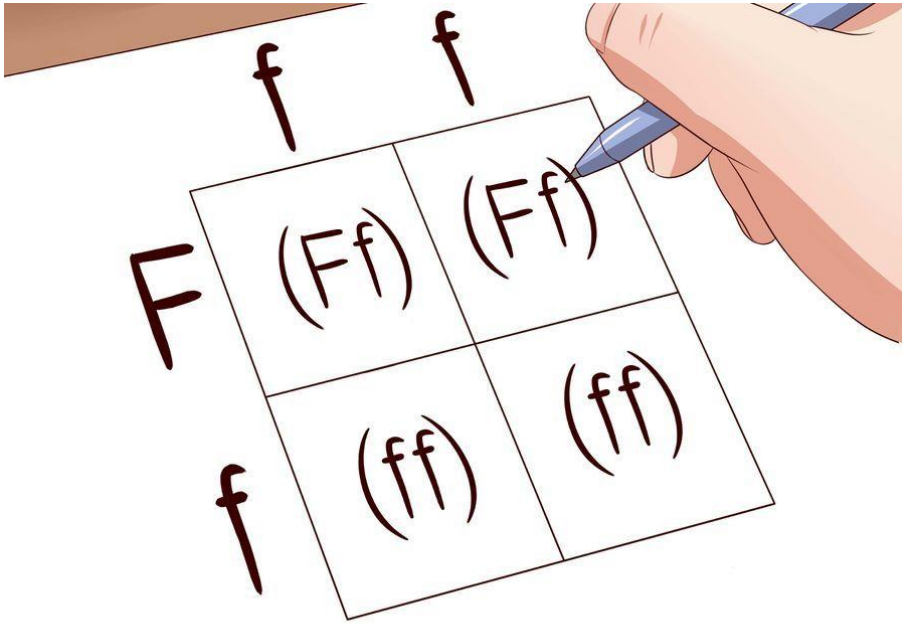
3. Write down all 3 possible genotypes and their letter combinations.

4. Select the correct genotypes for each parent (they may be the same or different) and write them with one parent on the top and one parent to the left side.



wikiHow to Make a Punnett Square

How to use a Punnett squares to predict offspring



wikiHow to Make a Punnett Square

5. Write each letter in the square below for one parent and squares to the right for the other parent. Each offspring will have a genotype of 2 letters.

6. Calculate the genotype ratio – the total percentage each genotype occurs (in this order $FF : Ff : ff$) or total out of 4 squares. Then calculate phenotype ratio – FF and Ff count as dominant and only ff counts as recessive (in this order dominant : recessive) can be % or out of 4



wikiHow to Make a Punnett Square

Using Punnett squares to predict offspring

The Punnett square is used to predict the **probability** of what the offspring's phenotype and genotype will be, which may or may not match up to the actual results due to the random nature of each fertilisation.

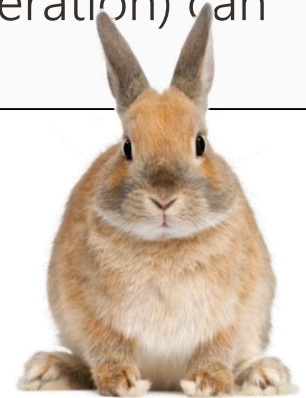


The phenotype and genotype ratios are only **predictions**. Each time a new offspring is created through fertilisation it is a new event and the same probabilities apply regardless of the phenotype or genotype of previous offspring

Calculating Phenotype and Genotype ratios

We can use the example of our straight eared and lop eared rabbit again when they breed, and all their offspring will have the genotype of Rr and phenotype of straight ears. If we cross two of their offspring (Rr) the genotype and phenotype ratios of **their offspring** (second generation) can be set out as below.

	R	r
R	RR	Rr
r	Rr	rr



Genotype Ratios		
RR	Rr	rr
1	2	1
Phenotype Ratios		
Straight Ears		Lop ears
3		1

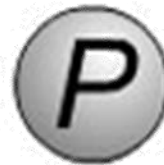
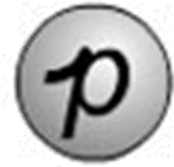
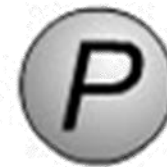
Phenotype and Genotype ratios




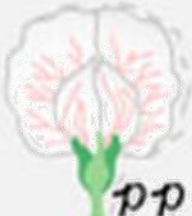
Genotype ratios when crossing two heterozygous parents are always 1:2:1.
That is 1 dominant homozygous : 2 heterozygous : 1 homozygous recessive. They are always written in that order
i.e. PP:Pp:pp

Genotype ratios when crossing one dominant homozygous and one recessive homozygous are always 0:4:0 with 100% of the offspring being heterozygous.

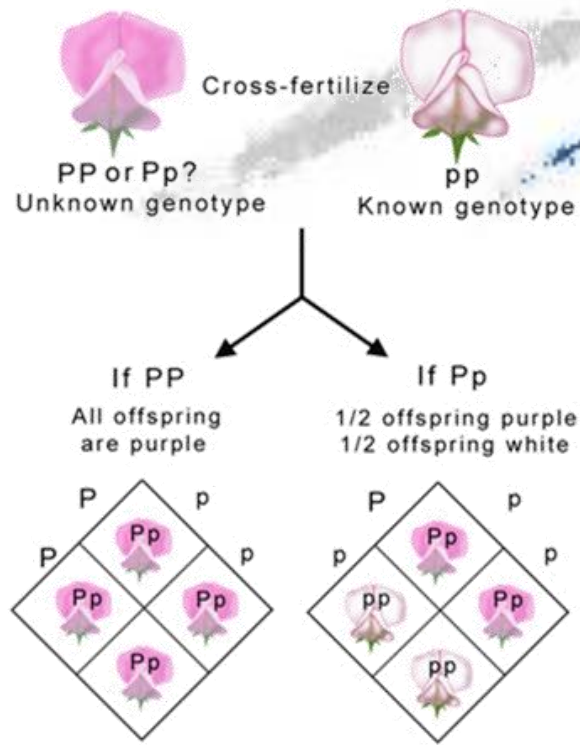
Phenotype ratios

Will always be written as dominant:recessive
Remember both PP and Pp will have the dominant phenotypes
and only pp will have the recessive phenotype.



 PP	 Pp
 Pp	 pp

Pure Breeding and test Crosses



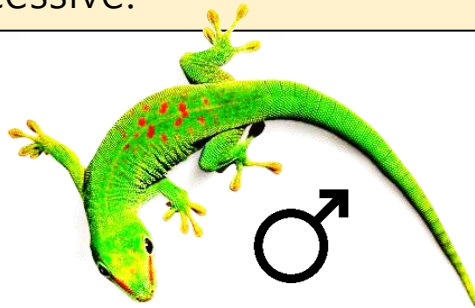
In order to establish a pure breeding population the **test cross must be repeated many times**. If the tested parent is not pure breeding then an estimated 50% of the offspring will show up with the recessive phenotype but a small sample size may not show this with just one cross. **Non-pure breeding heterozygous individuals must be continuously removed** from the population and not be allowed to breed further.

An individual that is **pure breeding** has a homozygous (two of each allele) genotype. The pure breeding recessive homozygous can be seen by its phenotype – it will show the recessive phenotype. The pure breeding dominant homozygous has the same phenotype as a heterozygous individual however. The pure breeding dominant individual can be found by a **test cross** which it mates with a recessive phenotype individual. If any of the offspring have the recessive phenotype then the individual is not pure breeding.

Test Crosses

The test cross is used to determine the genotype of an unknown individual that displays the dominant phenotype. The individual is crossed with a known homozygous recessive.

Genotype:
Unknown
RR or Rr
Phenotype:
green skin



X
Crossed with



Genotype:
Homozygous
recessive rr
Phenotype:
red skin

gametes	r ♂	r
R	R r	R r
♀ R	R r	R r

If the phenotypes are all Green skin then the unknown genotype is RR

Possible outcomes

Note: a Rr could also be used for test cross but instead of 50:50% green:red, the predicted will be 75:25%

gametes	r ♂	r
R	R r	R r
♀ r	r r	r r

If any of the phenotypes appear as red skin then the unknown genotype is Rr

Using Punnett squares to predict offspring

1. **Determine** the **genotypes** of the parents or whatever is given in problem.
2. Set up your **Punnett square** as follows:

Male – BB genotype
brown eyes -
phenotype

Female – bb genotype
blue eyes - phenotype

Genotypic ratio = 100% Bb

Phenotypic ratio = 100% Brown
eyes

	male	
	B	B
female	b	
	b	

3. **Fill in** the squares. This represents the possible combinations that could occur during fertilization.
4. **Write out** the possible **genotypic ratio** of the offspring.
5. **Using** the **genotypic** ratio determine the phenotypic ratio for the offspring.

Using Punnett squares to predict offspring

A heterozygous male, black eyed mouse is crossed with a red eyed, female mouse. Predict the possible offspring!

Male – Bb genotype
black eyes - phenotype

Female – bb genotype
red eyes - phenotype

Black must be dominant (B) as phenotype is black when mouse is heterozygous (has both B and b)

	male	
	B	b
female		
b		
b		

Genotypic ratio = 50% Bb 50% bb

Phenotypic ratio = 50% Black eyes 50% red eyes

Using Punnett squares to predict offspring

A heterozygous, smooth pea pod, plant is crossed with a wrinkled pea pod plant. *There are two alleles for pea pod, smooth and wrinkled.* Predict the offspring from this cross.

heterozygous Bb genotype
smooth - phenotype

homozygous bb genotype
wrinkled - phenotype

Smooth must be dominant (B) as phenotype is smooth when plant is heterozygous (has both B and b)

	smooth	
	B	b
wrinkled		
b		
b		

Genotypic ratio = 50% Bb 50% bb

Phenotypic ratio = 50% smooth 50% wrinkled

Using Punnett squares to predict offspring

Two heterozygous, smooth pea pod, plants are crossed. *There are two alleles for pea pods, smooth and wrinkled.* Predict the offspring from this cross.

heterozygous Bb genotype
smooth - phenotype

Smooth must be dominant (B) as phenotype is smooth when plant is heterozygous (has both B and b)

	smooth	
	B	b
wrinkled		
B		
b		

Genotypic ratio = 25% BB 50% Bb 25% bb
Phenotypic ratio = 75% smooth 25% wrinkled

Using Punnett squares to predict the Parent's genotype

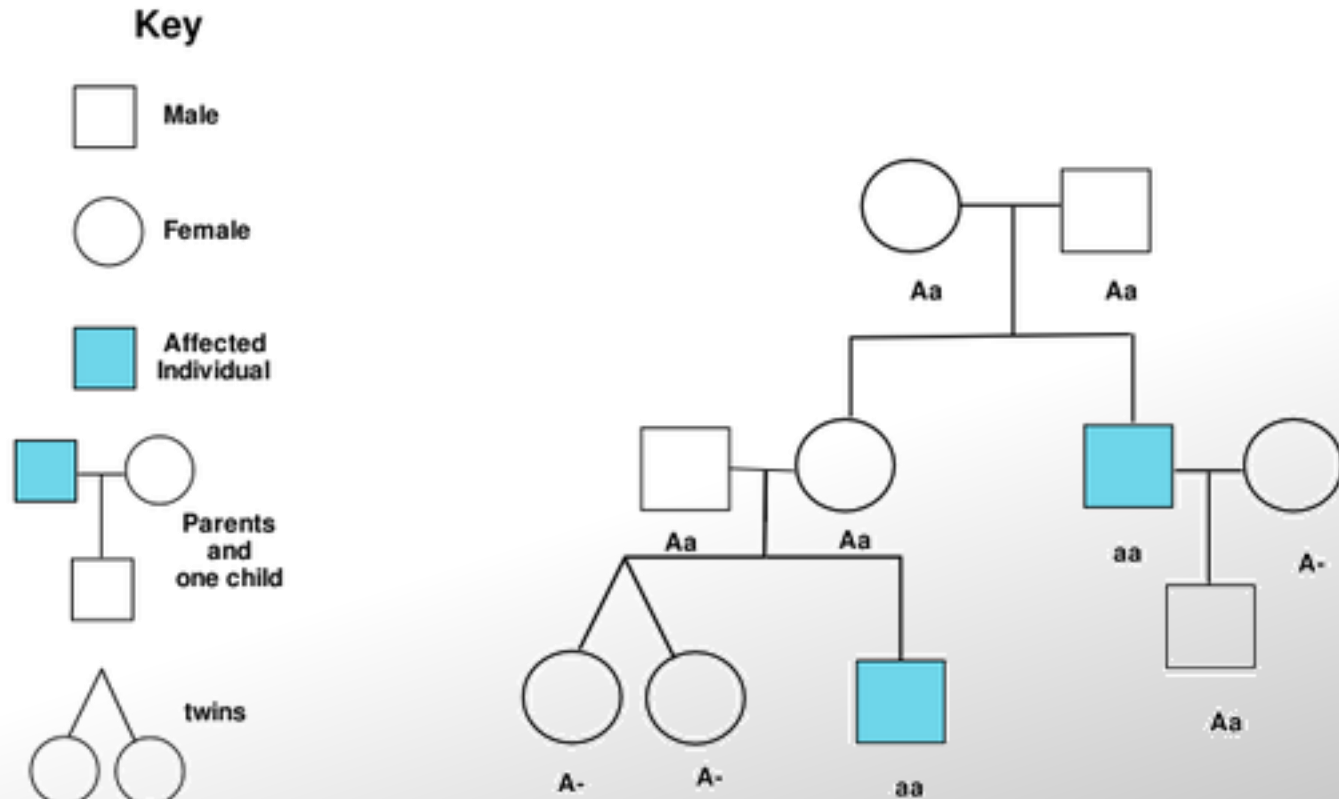
Parent's genotypes can be predicted by the phenotype of the offspring. If 100% of the offspring show the dominant phenotype then at least one of the parents must be homozygous dominant.

If any of the offspring show the recessive phenotype then each parent must contain at least one recessive allele each in order to have offspring that has a recessive allele donated from each parent. If the parents show the dominant phenotype then they must be heterozygous.

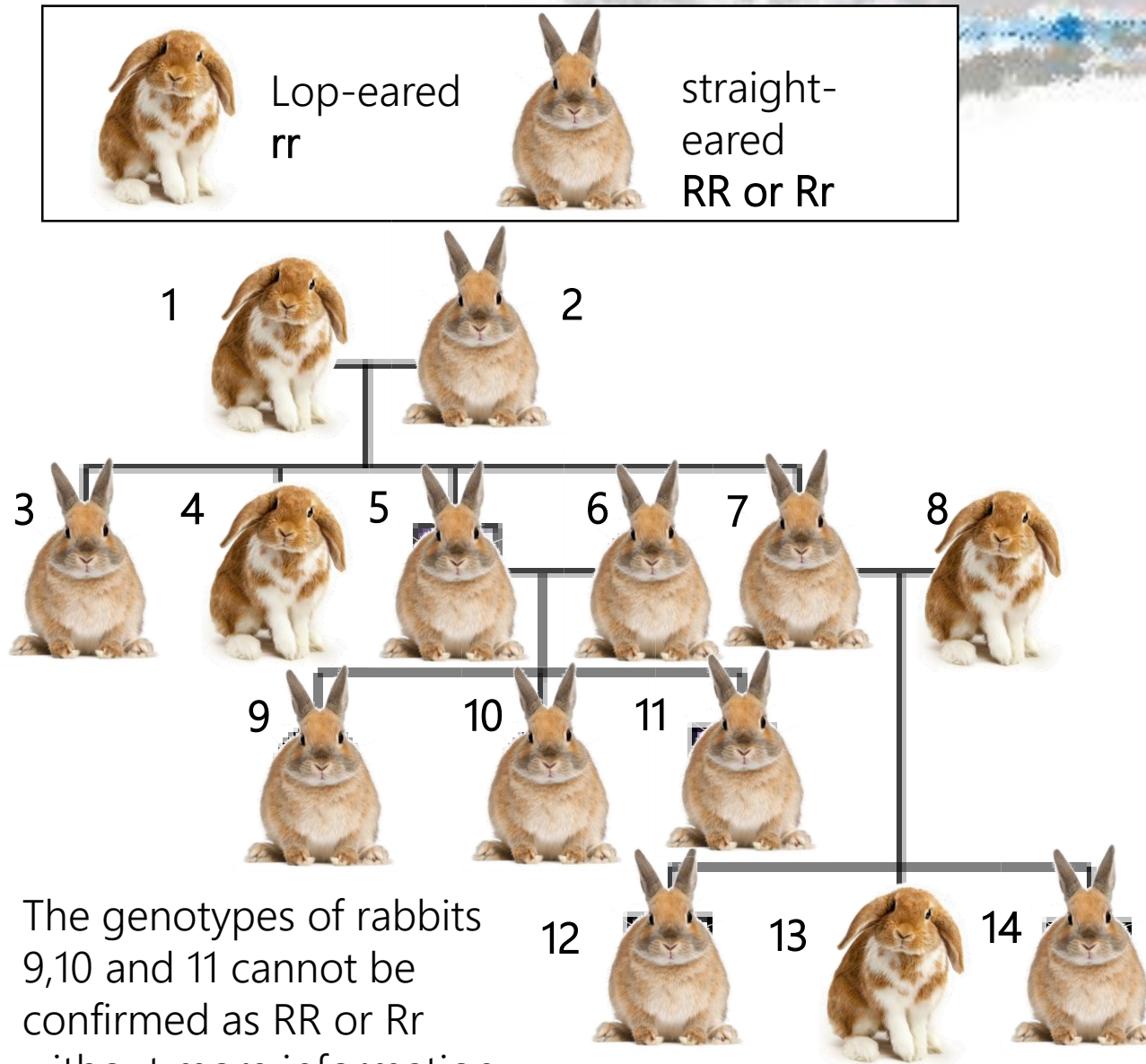


Pedigree charts

A pedigree chart is a diagram that shows **inheritance** patterns of a certain trait (Phenotype). A square represents a male and a circle represents a female. If a person's symbol is shaded in, this means that they have the phenotype (in this case, it is a recessive phenotype). If they are not shaded, they do not have the trait and their genotype can be either homozygous dominant or heterozygous. Pedigrees are good for showing the patterns of a recessive or dominant gene.



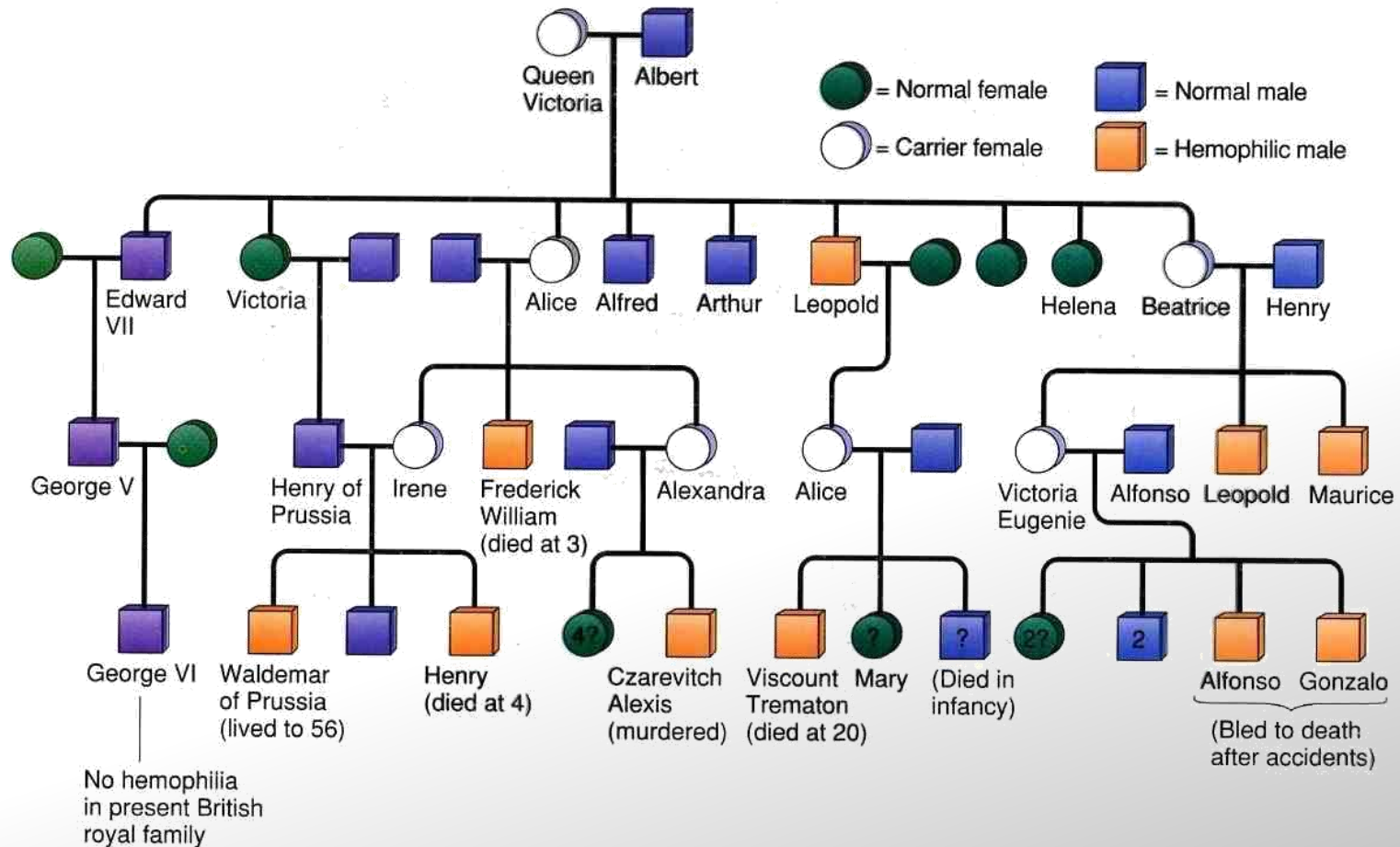
Using Pedigree charts to predict genotype



When an individual has a recessive phenotype on a pedigree chart then the **genotype must always be homozygous recessive** (i.e. rr for the lop-eared rabbit 1). To find out the genotype of a dominant phenotype you can look at either the parents of the offspring. (i.e. rabbit 7 is straight eared but had one parent who was a lop-eared rr so must have one r allele – genotype Rr . Also 13 is an rr (see above) so rabbit 7 must have at least one r allele to pass onto 13.

Using Pedigree charts to predict genetic disorders in offspring

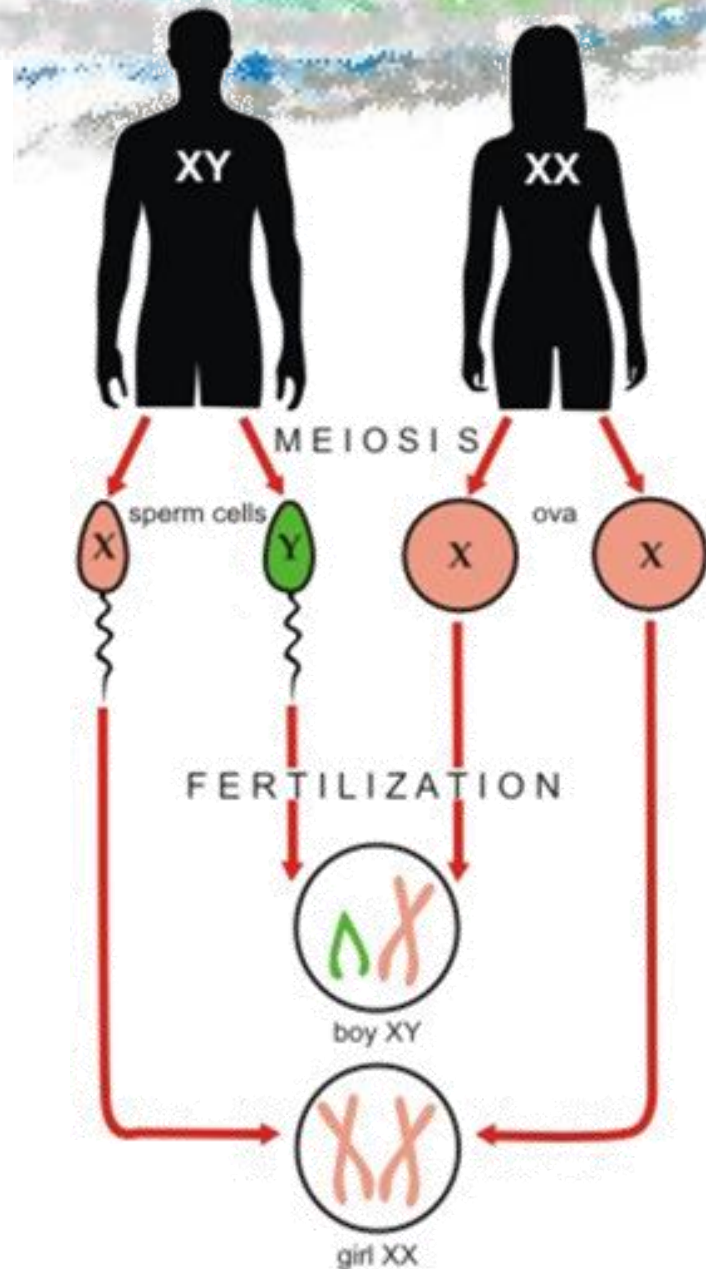
A pedigree chart can be used to predict the likelihood of offspring receiving a mutation from parents as well as look for where the mutation has come from. A particularly famous pedigree of a mutation in a family comes from the European royal family. The mutation is haemophilia (which prevents clotting of the blood) and can be traced from Queen Victoria



Sex determination

One of the 23 pairs of human chromosomes are called the **sex chromosomes**. They are either x or y. The female always has a pair of two x chromosomes. The female can only pass an x chromosome to their gametes (egg). The male has a x and y chromosome. He can pass either an x or y chromosome to form a gamete (sperm). The male determines the gender of any children.

Each offspring receives one copy of each chromosome from each of its parents during fertilisation, one from the sperm and one from the egg.



Using a Punnett square to show Sex determination

A Punnett square can be used to demonstrate that in any fertilisation there will be a 50% chance of either a boy or a girl.

If there are a small number of offspring then there is less chance that the actual ratio of male to female offspring will be the same as the predicted ratio.

Each new fertilisation is independent of any previous fertilisations'. Regardless of how many boys or girls have been produced previously in a family, each new child has a 50:50 of being a boy or girl.



		Male gametes	
		X	Y
Female gametes	X	XX	X ^Y
	X	XX	X ^Y

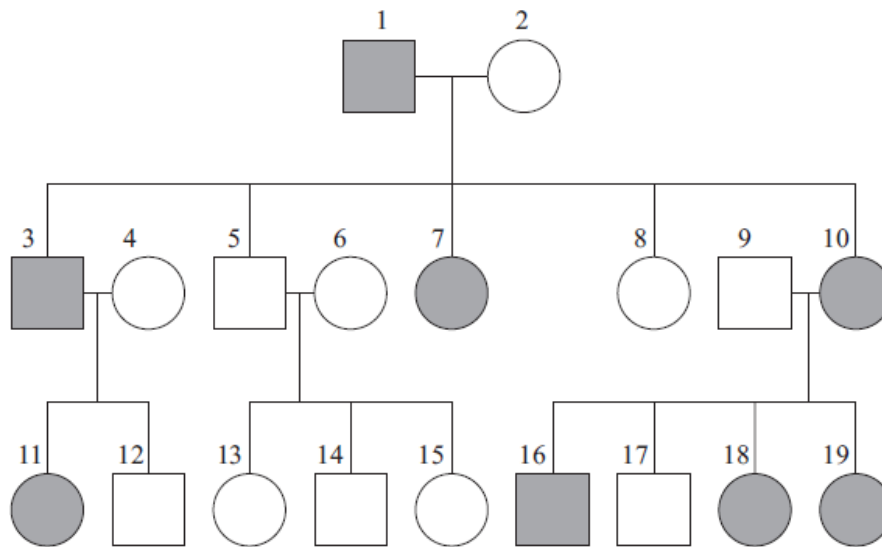
NCEA 2013 Inheritance - Huntington's disease

Achieved
Question

Question 1a: Huntington's disease is a genetic disorder in humans. It is caused by a dominant allele (H). The normal allele is recessive (h).


(a) Using H and h, give the two possible genotypes for an individual who has Huntington's disease:

Pedigree chart




affected male


affected female


unaffected male


unaffected female

Possible genotypes for an individual who has Huntington's are HH or Hh.

Question 1b: State the genotype of individual 9 in the pedigree chart above.

Genotype of parent 9 (male) is:
hh/homozygous

Achieved
Question

Question 1c: State the genotype of individual 10 in the pedigree chart above.

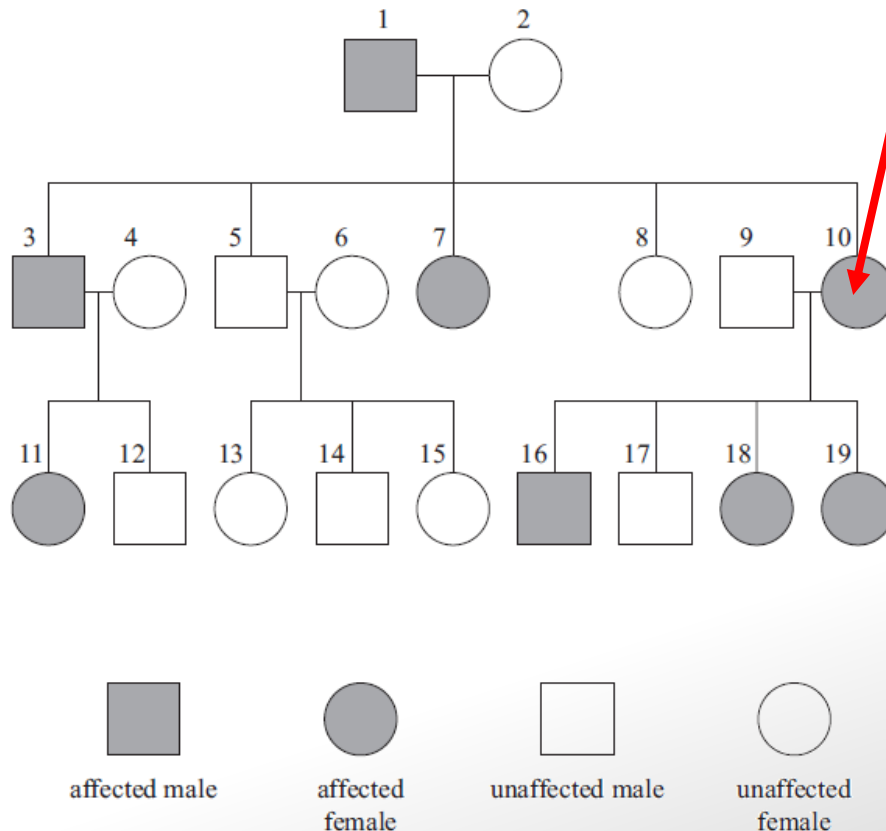
Genotype of parent 10 (female) is:
Hh/heterozygous

NCEA 2013 Inheritance - Huntington's disease

Excellence
Question

Question 1b(iii): Explain how you worked out the genotype for individual 10. You should support your answer using evidence from BOTH the parents AND children of individual 10.

Pedigree chart



Genotype of parent 10 (female) is:
Hh/heterozygous

The females parents are Numbers 1 and 2. Her mother (number 2) is unaffected but her father (number 1) has Huntington's disease. Number 2 is hh and number 1 must have at least one dominant allele. Because one allele is inherited from each, parent No. 2 must have given her daughter a recessive allele. She has inherited the dominant allele from her father, No. 1. She cannot be homozygous dominant HH because one of her offspring, No. 17, is not affected by the disease and is therefore hh.

NCEA 2013 Inheritance - Huntington's disease

Achieved
Question

Question 1c: Draw a Punnett square to show the **possible** genotypes of the children from parents 9 and 10. From **your** Punnett square, predict what fraction of the children would have Huntington's disease and what fraction would not have Huntington's disease.

Genotype of parent 9 (male) is: hh/homozygous
Genotype of parent 10 (female) is: Hh/heterozygous

		parent 10	
		H	h
parent 9	h	Hh	hh
	h	Hh	hh

Fraction of children with Huntington's disease is $\frac{1}{2}$
Fraction of children without Huntington's disease is $\frac{1}{2}$

Phenotype ratio is 1:1.

NCEA 2013 Inheritance Predictions - Huntington's disease

Excellence
Question

Question 1d: In the pedigree chart the phenotype ratio of Huntington's disease in the children of parents 9 and 10 is not the same as the predicted ratio in the Punnett square

Give reasons why the predicted ratio in the Punnett square and the observed ratio in the children may NOT be the same.

parent 9

parent 10		
	H	h
h	Hh	hh
h	Hh	hh

Fraction of children with Huntington's disease is $\frac{1}{2}$
 Fraction of children without Huntington's disease is $\frac{1}{2}$
 Phenotype ratio is 1:1.

Answer 1d: Punnett squares **predict** probable offspring genotypes (thence the expected phenotypes) based on the gametes of the parents. Pedigree charts give the observed (actual) phenotypes. Since each fertilisation is a random event, it is by chance whether the offspring of number 10 inherits the dominant H allele and therefore has Huntington's or the recessive h allele and does not have the disease. In the pedigree chart 3 of the 4 offspring have the disease but only 2 out of 4 would have been predicted from the Punnett square.

NCEA 2013 Inheritance – Eye Colour

Excellence
Question



	B	b
B	BB	Bb
b	Bb	bb

Question 2b: The allele for brown eyes (B) is dominant over the allele for blue eyes (b) in humans.

Discuss how it would be possible for a child to have blue eyes, even though both their parents have brown eyes. In your answer you should:

- use labelled Punnett squares
- link the genotypes and phenotypes of the child, parents, AND grandparents.

Answer 2b: For the child to have blue eyes they must have a genotype of bb (ie have both recessive alleles). If a dominant allele, B is present then brown eyes would be seen. In order to have a genotype of bb, each parent must have given a b (recessive allele). Both parents have brown eyes so therefore they both must have a dominant allele (B) and because each parent passes on a recessive allele the genotype of each parent must be Bb. The grandparents could have a genotype of bb, Bb, or BB. It is not possible to say for sure, but at least one of the grandparents on each side must pass on a recessive allele (b) in order for each parent to have a recessive allele to pass on to the child. Punnett square(s) may be used to show this but must be explained.

NCEA 2014 Inheritance – sheep wool colour

Question 3a: An animal breeder wanted to produce sheep with white wool, but some white sheep produce lambs that have black wool. Animal breeders often use one male sheep to mate with all their female sheep. Give all possible genotypes for each phenotype. Use **A** to represent the dominant allele for common white wool, and **a** to represent the recessive allele for black wool.



A white sheep could have genotype AA or Aa, whereas a black sheep can be only aa.

NCEA 2014 Inheritance Predictions - sheep wool colour

Excellence
Question

Question 3b: Discuss how a farmer could develop a group of sheep that are pure breeding for white wool.

In your answer you should:

- state the genotypes of the male and female sheep the farmer should use to breed from
- explain how the animal breeder can determine the genotypes of the male and female to produce sheep that all have white wool.

You should include at least two Punnett squares with your explanation

- explain how the animal breeder could make sure that the offspring would always be pure breeding.

For a test cross, a large / reasonable number of offspring need to be produced to be sure a sheep is AA.

AND as soon as a black offspring appears, the breeder knows the white sheep is Aa.

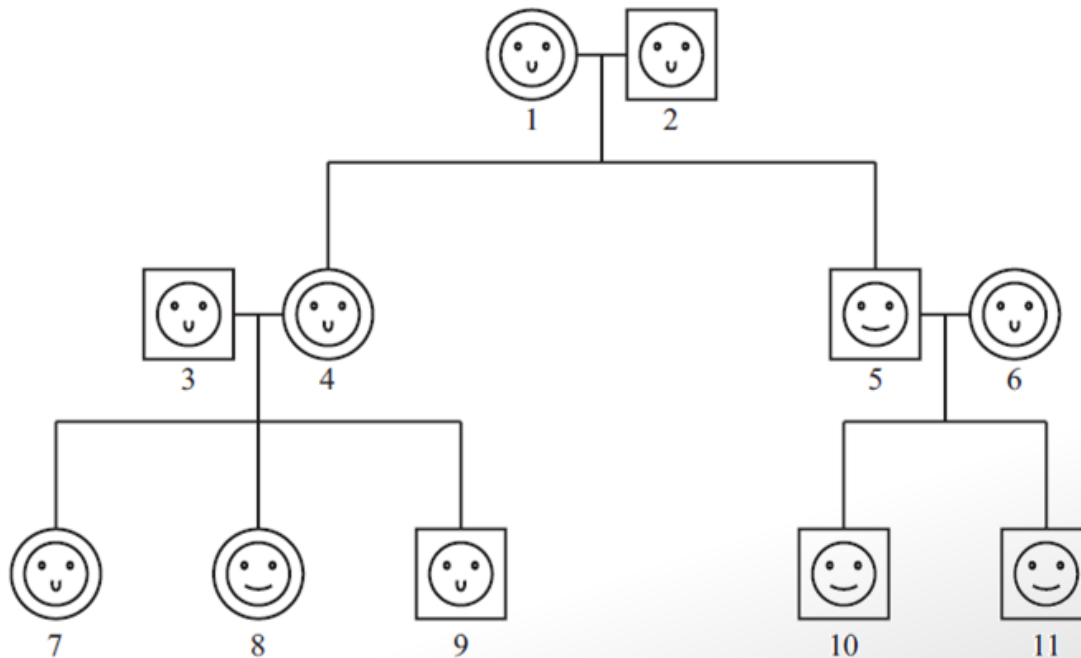
Answer 3c: To breed a group of white sheep, a breeder should use sheep that are both AA. The breeder can determine if a white sheep is AA by crossing a white sheep with a black sheep. If the white sheep is AA, none of the offspring will be black. The breeder would need to carry out many crosses to show that it was not just due to chance that a black sheep had not been produced. If a black offspring is produced, the breeder can be certain the white parent was Aa. The farmer should breed only with an AA male and white wool females, as this will ensure that all offspring are white. Ideally, the breeder would breed from AA males and AA females, as this would remove the recessive allele from the group.

NCEA 2014 Inheritance – Tongue rollers

Achieved
Question

Question 4a: In the family tree below, people who are tongue rollers are shown as: ☺ while those who cannot roll their tongue are shown as: ☹
Use the letters **T** and **t** to represent the alleles for tongue rolling (**T**) and non rolling (**t**).

- (a) (i) Use the family tree above to work out the genotype of individual 5.
(ii) Explain how you worked this out.



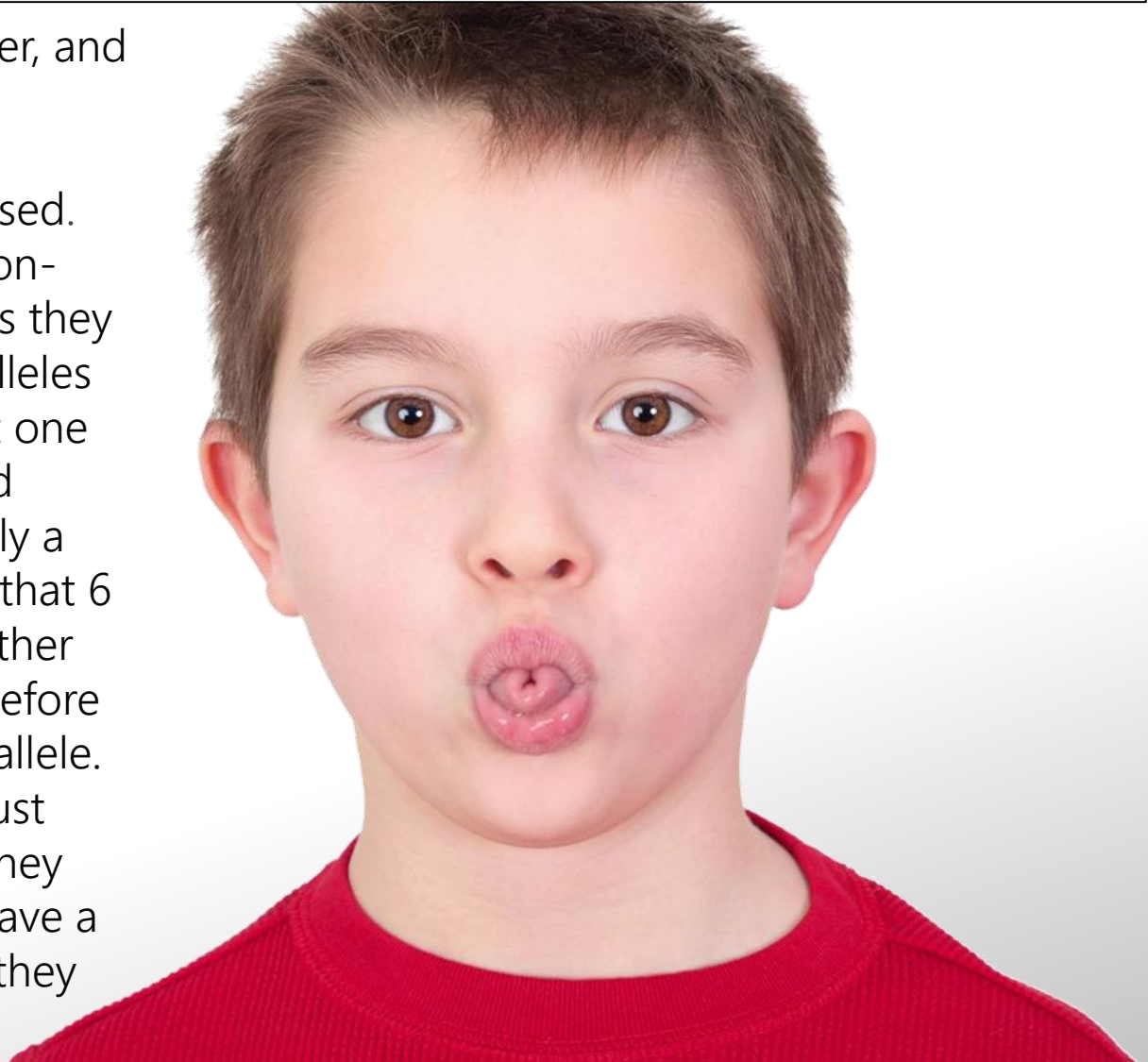
Genotype individual 5 =
 tt

If a dominant allele was present, then individual 5 would be a tongue roller. So therefore they must have only both recessive alleles present.

NCEA 2014 Inheritance – Tongue rollers

Question 4b: Use the family tree to explain why individual 6 must be Tt.

Individual 6 is a tongue roller, and so must have at least one dominant allele present for tongue rolling to be expressed. Both of 6's children are a non-tongue rollers, which means they must have both recessive alleles and be tt. Because they get one allele from each parent, and individual 5 can pass on only a recessive allele, this means that 6 must have passed on the other recessive allele, and so therefore 6 must have one recessive allele. Therefore, because they must have a dominant allele so they can tongue roll and must have a recessive allele to pass on, they must have genotype Tt.



NCEA 2014 Inheritance – Tongue rollers

Question 4c: Explain why the genotypes for individuals 3 and 4 both must be **Tt**.

In your answer you should:

- draw Punnett squares in the box below
- explain why the genotypes of individuals 3 and 4 **cannot** be **TT** or **tt**.

Individuals 3 and 4 cannot be **tt**, as they are both tongue rollers. For them both to be tongue rollers, each of them must have at least one dominant allele, so this precludes **tt** as a possible genotype. It is not possible for individuals 3 and 4 to be **TT**, as one of their offspring is a non-tongue roller. A non-tongue rolling child must have genotype **tt**, because if they have a dominant allele at all, they would be able to tongue roll. One of each allele must come from each parent, and so for the child (individual 8) to be **tt**, a recessive allele must have come from each parent. For this to occur, each parent must have a recessive allele. Because the parents are both tongue rollers, they must each have a dominant allele, and because they both must have a recessive allele to pass on, they can only be **Tt**.

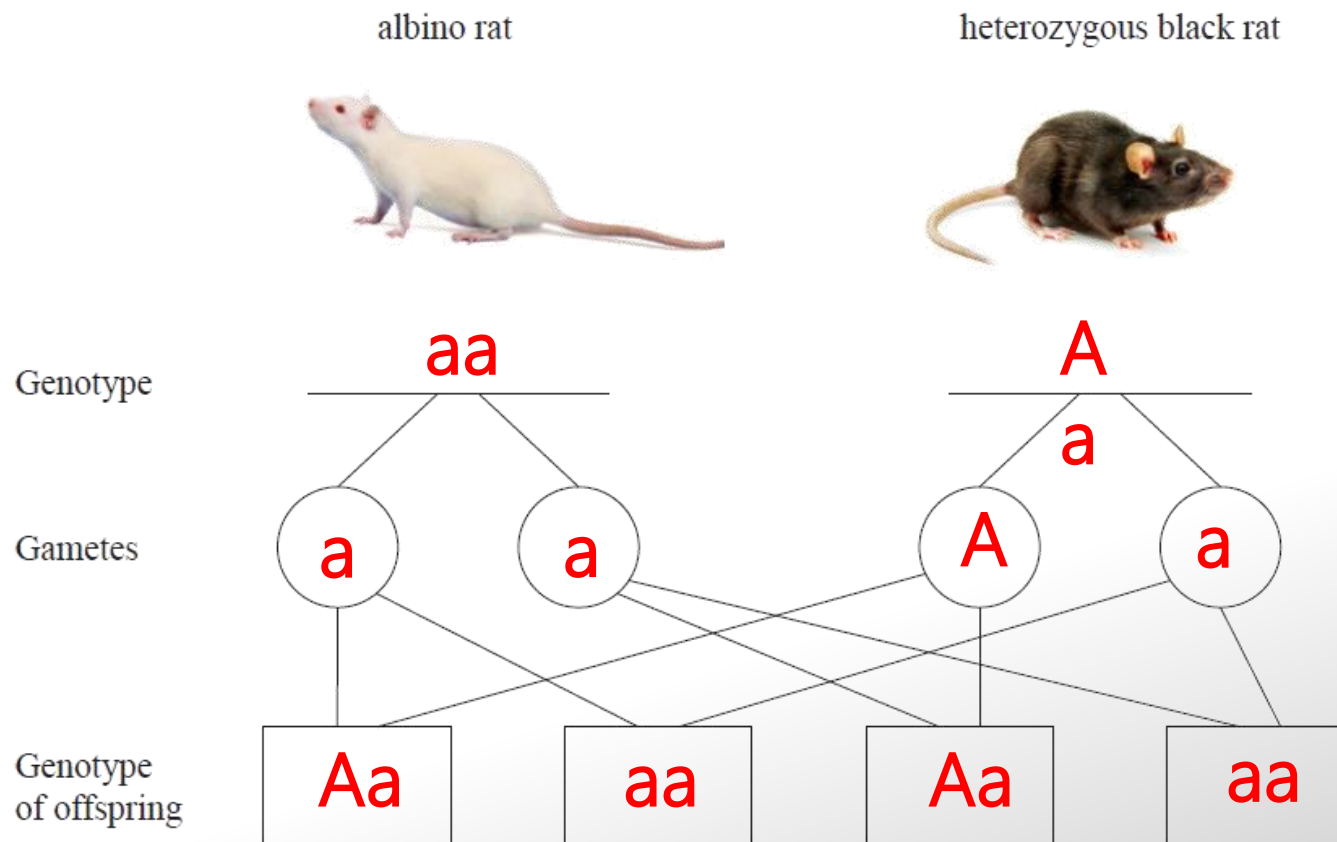
Relevant Punnett squares drawn which could be:

A cross showing the two heterozygous parents.

A cross showing individual 3 or 4 with genotype **TT** and therefore shows that they cannot be **TT**.

NCEA 2015 Inheritance – Albino Rats (PART ONE)

Question 1a: Albinism in rats results in white fur and pink eyes. Albinism is caused by a recessive allele **a**. Complete the following diagram:



Achieved
Question

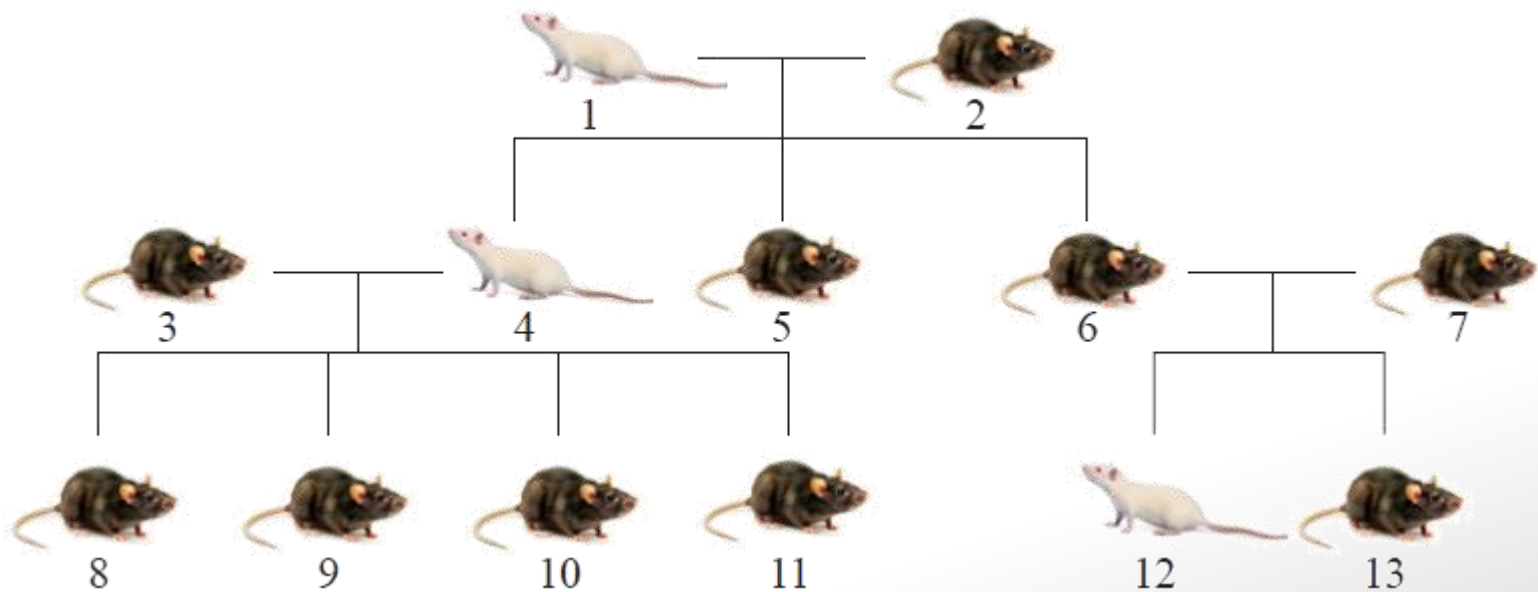
Achieved
Question

NCEA 2015 Inheritance - Albino Rats (PART TWO)

Achieved
Question

Question 1b: The albino rat (1) and the heterozygous black rat(2) produced the following two generations of offspring, as shown in the pedigree chart below.

What are the genotypes of the following rats? Rat 4, Rat 6, Rat 10



Answer 1b:

Rat 4 aa/ homozygous recessive

Rat 6 Aa /heterozygous

Rat 10 Aa /heterozygous

NCEA 2015 Inheritance - Albino Rats (PART THREE)

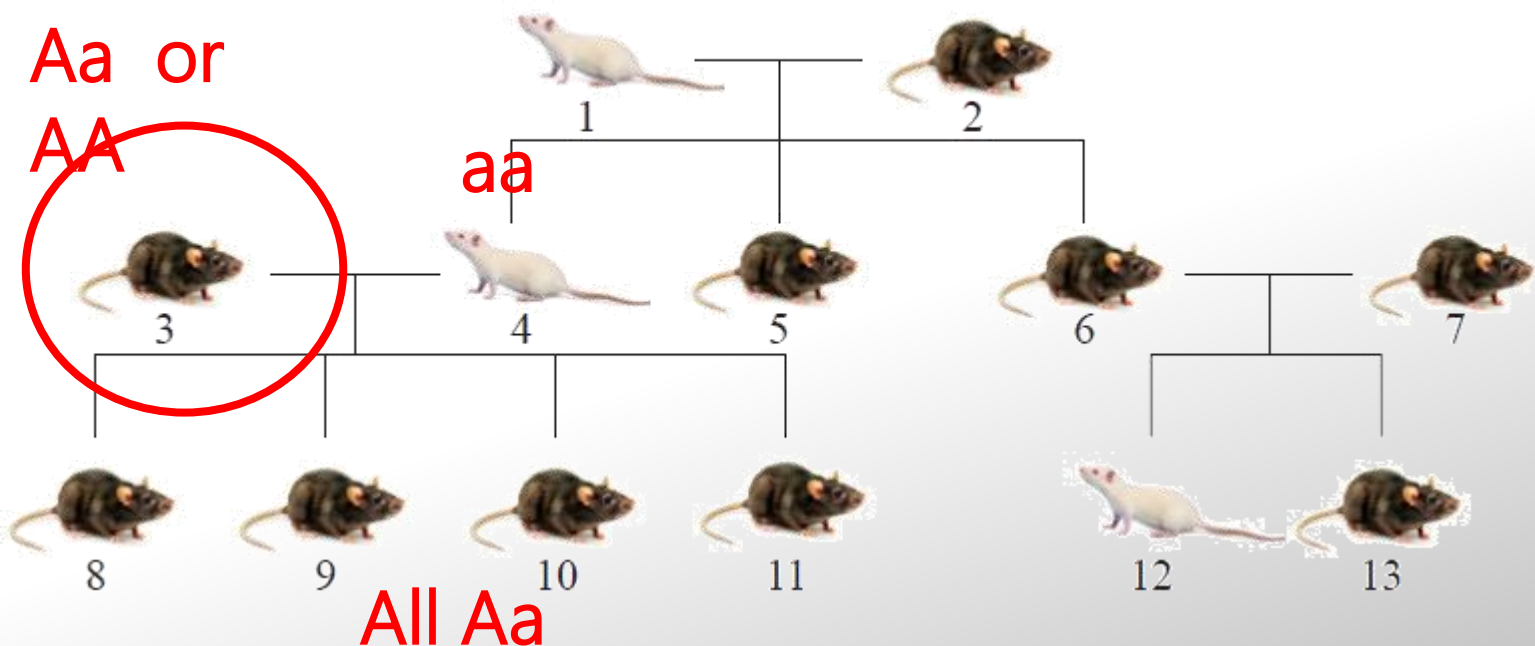
Excellence
Question

Question 1c: Rat 3 was **not** an offspring of Rat 1 and Rat 2 in the family tree.

Give the possible genotypes for Rat 3 and explain which is the most likely genotype for Rat 3.

In your answer you should:

- state the possible genotypes for Rat 3
- explain why both genotypes are possible but one is more likely
- explain what you could do to be more certain about the genotype of Rat 3.



NCEA 2015 Inheritance - Albino Rats (PART THREE)

3
Excellence
Answers**Answer 1c:**

The black Rat 3 must have at least 1 dominant A allele because its phenotype is black. However there are two possible genotypes **AA or Aa**. These genotypes could result in the following crosses:

	A	A		A	a
a	Aa	Aa	a	Aa	aa
a	Aa	Aa	a	Aa	aa

The pedigree tree provided shows that all the actual offspring were black; therefore the most likely genotype for Rat 3 is AA, as this can only produce black offspring. However these Punnet squares only show the probability of an event occurring. The Aa / aa cross can also produce black offspring. It may just be chance that aa offspring were not produced. To be certain of the genotype of the black Rat 3, you would have to carry out many more crosses with the aa parent. If, after a **LARGE number of crosses**, there were no white offspring you would have confidence the rat was AA, but only one white offspring will prove Aa.

The Rat 3 is **more likely** to be AA, because ALL the offspring from crossing individuals 3 and 4 were black.

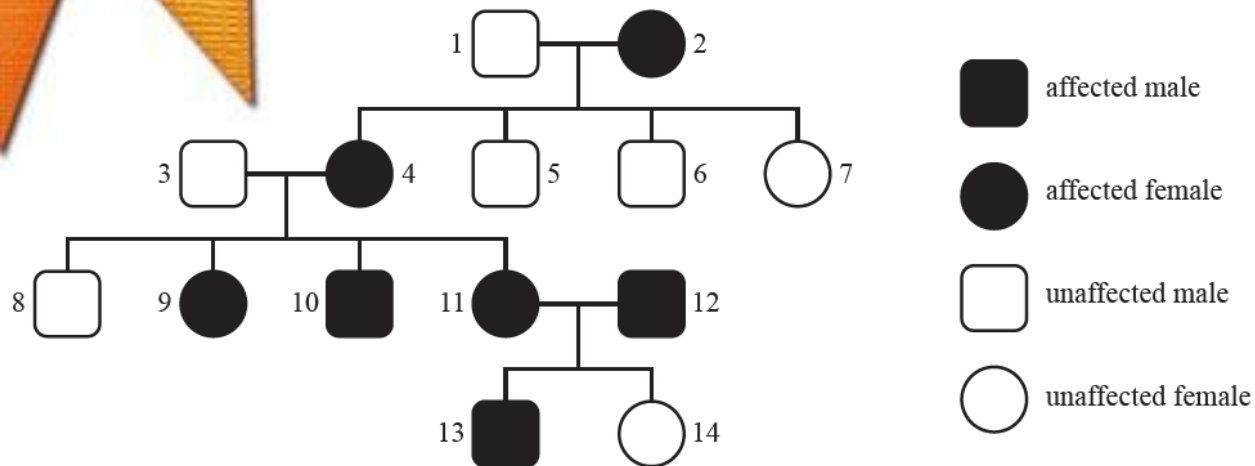
Although AA is **more likely** / probable, it is still possible that the black parent was Aa, and by chance aa was not produced.

After **many** crosses, if no white offspring occurred, you would have **more** confidence that black Rat 3 parent was AA AND if any (one) white offspring were produced that would prove Rat 3 was heterozygous Aa.

NCEA 2016 Inheritance – photic sneezing

Question 1a: Photic sneezing is a condition which causes affected people to sneeze due to bright light. It can be traced through a family, as shown in the pedigree chart. Photic sneezing (A) is dominant to unaffected (a).

(a) Work out the genotypes of the following four individuals: 1, 2, 11, 12



For a dominant trait any individual not showing the trait will have to be homozygous recessive. If one parent is recessive homozygous then all offspring will have at least one recessive allele

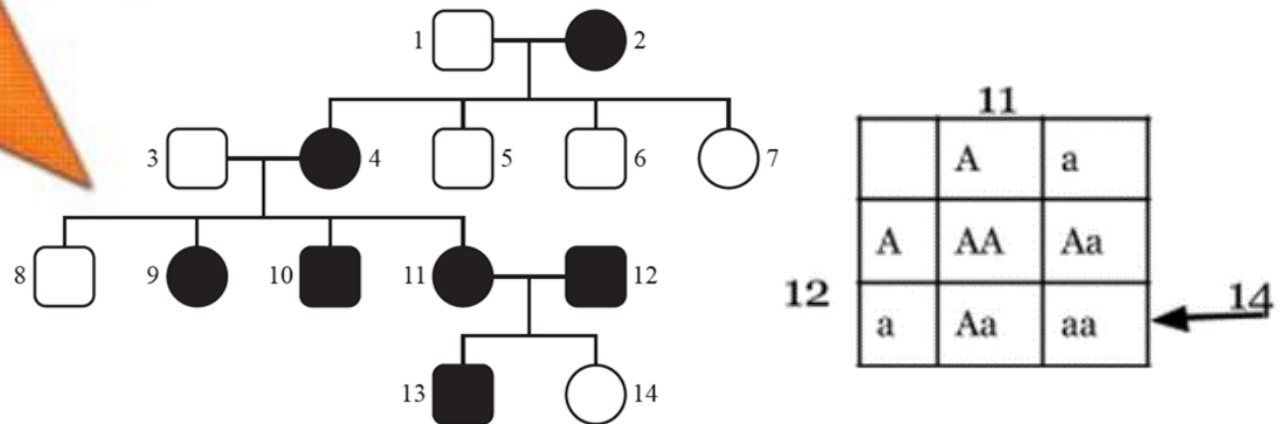
Answer 1a:

1 = aa, 2 = Aa, 11 = Aa, 12 = Aa

NCEA 2016 Inheritance – photic sneezing

Question 1b: Explain how the pedigree chart can be used to show that Photic sneezing is dominant, but it cannot be used to determine the genotype of individual 13.

You may use the Punnett square.



Answer 1b:

14 is a non-sneezing, but their parents are sneezers. The non-sneezing allele must be hidden in 11 and 12. Alleles that can be hidden are recessive.

OR If non-sneezing was dominant, individual 14 would have to have A. This must have come from one of their parents, who would therefore show the trait. But neither does, so non-sneezing cannot be dominant.

13 is a sneezer so must have A. They could be AA or Aa as each parent can pass on either. The Punnett square shows that 25% are expected to be AA and 50% Aa.

Without offspring we can't prove either.

NCEA 2016 Inheritance Predictions – photic sneezing

Question 1c: The cross between 1 and 2 in the pedigree chart has **one affected sneezing** offspring.

The cross between 3 and 4 in the pedigree chart has **three affected sneezing** offspring.

Explain the difference in the number of affected offspring (photic sneezers) in these 2 crosses.

In your answer you should:

- complete Punnett squares
- give the expected phenotype ratio for each cross
- account for any difference between the expected ratio and the actual phenotype ratio for each of the crosses.

Answer 1c:

Punnett square shows 50% sneezers are expected.

1 and 2 have 1, less than expected, while 3 and 4 have 3, more than expected. This is because each of the offspring is an **independent event**. Each offspring is **unaffected by previous outcomes**, and so each has a 50% chance of inheriting the a allele from #2.

We would expect to be very **close to 50% with a larger number of offspring**, e.g. if the offspring from 1 / 2 and 3 / 4 are combined it is 50%.

	1		
	a	a	
2	A	Aa	← 5,6,7
	a	aa	
			← 4

Question

NCEA 2017 Inheritance– Piebald Horses (Part ONE)

Question 2a: Piebaldism is a genetic condition causing a white patch on the head and body of horses. In horses piebaldism is a **dominant** trait (H), and “normal” colour is recessive (h).

<http://www.nzqa.govt.nz>

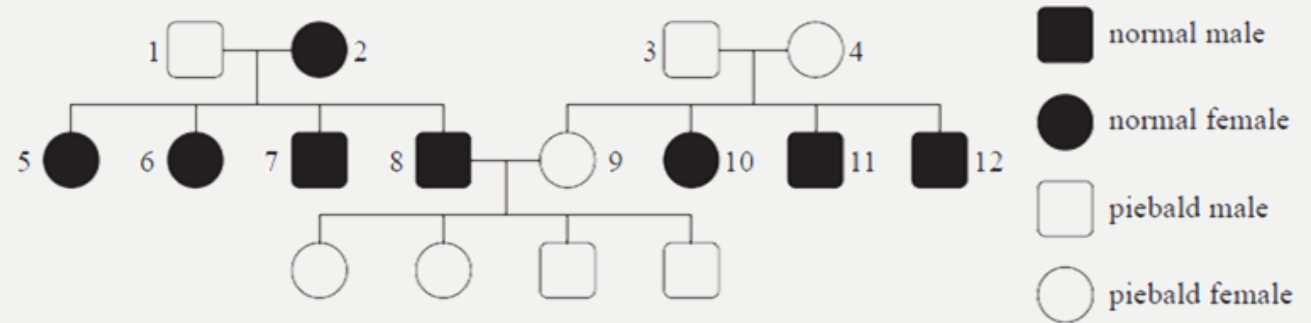


<https://www.pinterest.nz/pin/255297872600783620/>



<http://www.mybligr.com/beautiful-photographs-black-horse-20-pics/beautiful-black-horse-images-pictures-photos-13/>

Question 2a:



From the pedigree chart above, list **all** the possible phenotypes and genotypes of horses 3, 8, and 9.

Use the letter H for the dominant trait and h for the recessive.

individual	Phenotype (normal or piebald)	Genotype (HH,Hh, or hh)
3	piebald	Hh
8	normal	hh
9	piebald	HH or Hh

NCEA 2017 Inheritance– Piebald Horses (Part THREE)

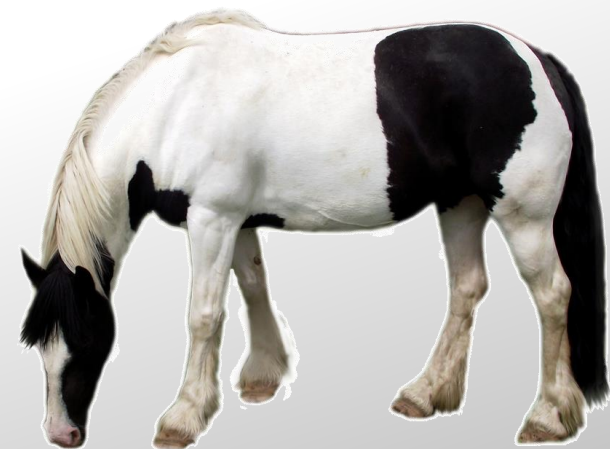
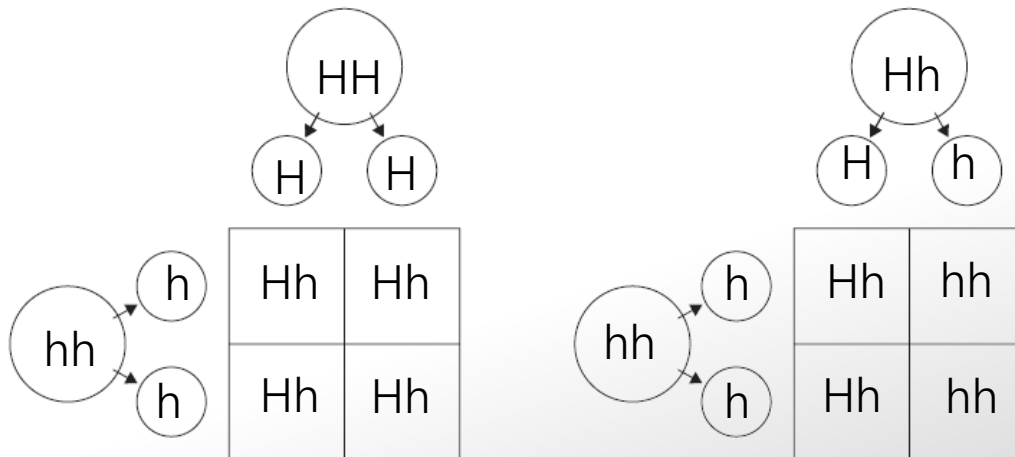
Excellence
Question

Question 2b: A breeder wants to produce only **dominant** (piebald) offspring from a breeding pair of horses. The breeder has piebald and normal horses to breed from. How could the breeder use crosses to make sure that the pair of horses were **pure breeding**? *Show crosses using Punnett squares to support your answer.*

<http://www.nzqa.govt.nz>

Pure-breeding individuals do not have any hidden information; they are homozygous. Piebald horses could be HH or Hh. Pure-breeding animals would have to be HH. To ensure that they were pure-breeding, the breeder could cross the horses with normal animals (hh).

If any of the offspring are normal, the piebald parent must be heterozygous – this cannot be used for breeding. If all the offspring are piebald she can start to be confident that the horse is a pure-bred. Many offspring are needed to be confident of the genotype but this cannot be proven.



<https://bimbobabe.deviantart.com/art/PRECUT-Piebald-horse-252473141>

NCEA 2018 Inheritance – Chicken Combs

Excellence
Question

<http://www.nzqa.govt.nz>

Question 1a: The allele for rose comb (R) is **dominant** to the allele for single comb (r) in chickens. (a) Two rose comb chickens produce a single comb offspring. Explain how it is possible for two rose comb chickens to produce a single comb offspring. In your answer you should:

- define dominant allele
- explain the genotypes of the parents and offspring
- use a Punnett square to help your explanation.

Dominant alleles are always expressed if present, whereas recessive alleles can be hidden (by a dominant allele). The single comb offspring must be rr to show the recessive phenotype. Both of the parents have to have a dominant allele (R), as they have rose combs. They must also have the recessive allele (r) to be able to pass on an r to the rr offspring. In this way, the hidden allele (single) can be passed on from the parents to show up in the offspring.

	R	r
R	RR	Rr
r	Rr	rr



<https://www.flickr.com/photos/archer10/7815488864>
Rose comb on a chicken



<https://pixabay.com/en/hahn-cockscomb-comb-teeth-farm-66341/>

Single comb on a chicken

NCEA 2018 Inheritance – Test Cross

Excellence
Question

<http://www.nzqa.govt.nz>

Question 1b: Explain how a breeder could use crosses to find out if a rose comb chicken has a pure breeding genotype for the trait.

In your answer:

- define pure breeding and genotype
- use Punnett squares to help you explain
- explain when the breeder could be confident of the chicken's genotype.

Pure breeding means homozygous for the rose comb trait / RR. Genotype means the alleles present for a trait. A rose comb chicken must be either RR or Rr. To find if it has a recessive allele, it should be crossed with a single comb chicken (rr). [**or a Rr chicken*]

An RR × rr cross will give 100% rose comb, whereas an Rr × rr cross will give 50% rose and 50% single. [**or equivalent for RR × Rr (0%) and Rr × Rr (25%).*]

If **any** of the offspring are single combed, the rose comb parent must be Rr. If all the **many** offspring are rose combed, the parent is **likely** to be RR (pure breeding).

	R	r
r	Rr	rr
r	Rr	rr

	R	R
r	Rr	Rr
r	Rr	Rr



Question 3b: Explain how the sexual reproduction of kauri trees causes genetic variation AND how this could lead to increased survival of the species when faced with kauri dieback disease.

In your answer you should consider:

- the processes of gamete formation (meiosis) and fertilisation
- how sexual reproduction leads to variation in the population
- the link between genetic variation and the survival of kauri trees as a species.

<http://www.nzqa.govt.nz>

Gamete formation by meiosis: Random assortment/segregation / crossing of chromosomes over in meiosis

Description of meiosis: produces gametes / sex cells that have half the normal number of chromosomes as body cells.

Process of fertilisation: Random male and female gametes join, each with unique DNA producing a genetically unique zygote / offspring.

Role of sexual reproduction: produce new combinations of alleles and thus genetic variation between individuals.

Explanation: The advantage of genetic variation to a species is that it may enable some individuals to survive kauri dieback to reproduce, passing on favourable alleles / genes to the next generation. Over many generations this genetic advantage / genes / alleles will rise in the population, allowing survival of the kauri species.



<http://www.nzqa.govt.nz>

Question 2c: Explain any difference in the expected phenotypes of the offspring ratio to the actual phenotypes of the offspring ratio between the cross with individual 1 and individual 2. In your answer you should refer to the expected and actual phenotype ratios for the cross.

Expected ratio 3:1 unaffected to CF respectively
(accept 75% unaffected to 25% CF or 3/4 unaffected and 1/4 affected).

Actual ratio 2:2 or 1:1 Unaffected to CF respectively
(accept 50% or 1 / 2 unaffected to 50% or 1 / 2 CF).

I-1 and I-2 have more CF than expected. This is because each offspring is the product of a random event/fertilisation.

Each offspring is unaffected by previous outcomes. Since I-1 and I-2 are both heterozygous (Tt), each individual has 50% chance of inheriting either allele from each parent.

With a larger number of offspring, we would expect a ratio very close to 3:1 unaffected to CF respectively.

