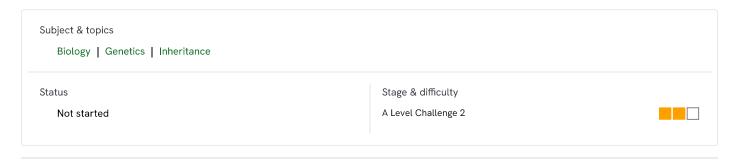


Agouti Genetics and Hair Colour



The *agouti* gene in mammals codes for a signalling protein that regulates the production of different types of melanin. There are two main types of melanin in mammals that are involved in hair pigmentation: *eumelanin* (which has a brown/black colour) and *pheomelanin* (which has a yellow/red colour). Because the agouti protein regulates where and when these are produced, different alleles of the *agouti* gene can result in different hair colour phenotypes.

The agouti gene is located on an autosome, and so males and females each have two copies of this gene.

Part A **Horses**

In horses, there are two main alleles of the agouti gene.

- The dominant allele, **A**, restricts eumelanin production to the mane, tail, and legs. Horses with this phenotype are mostly reddish-brown, but with a black mane, tail, and legs. This phenotype is called the Bay phenotype, and these horses are called *Bay horses*.
- The recessive allele, **a**, causes eumelanin production all over the body. Horses with this phenotype are black.

Two horses that are heterozygous for the agouti gene mate with each other.

Calculate the probability that the foal (young horse) has the Bay phenotype.

Part B

Cats

In cats, there are also two main alleles of the agouti gene.

- The dominant allele, **A**, causes 'agouti colouration', which is when individual hairs have alternating bands of eumelanin and pheomelanin. Cats with this phenotype are called *tabby cats*, though the exact type of tabby patterning depends on other genes.
- The recessive allele, **a**, causes hair to only contain eumelanin. Cats with this phenotype are black.

A male tabby	cat that is heterozygous	for the <i>agouti</i> gene	e mates with a	a female black	cat. The female	produces a litter
of 8 kittens.						

What is the expected number of black kittens in this litter?

Part C

Rabbits

In rabbits, there are three main alleles of the agouti gene: A, a^t, and a.

- The A allele is dominant to the other two alleles. Like in cats, this allele causes 'agouti colouration' (individual hairs have alternating bands of eumelanin and pheomelanin). Rabbits with this phenotype have a greyish-brown appearance, as is most commonly seen in wild rabbits, and are sometimes called agouti rabbits.
- The **a**^t allele is recessive to the **A** allele and dominant to the **a** allele. This allele causes eumelanin production everywhere except the underside, and so rabbits with this phenotype are black with a white belly. Rabbits with this phenotype are sometimes called *otter rabbits*.
- The **a** allele is recessive to the other two alleles. This allele causes eumelanin production all over the body. Rabbits with this phenotype are black.

A breeder performs a cross (cross 1) between a homozygous agouti rabbit and a black rabbit. They perform a separate cross (cross 2) between a homozygous otter rabbit and a black rabbit.

Finally, they perform another cross (cross 3) between one of the males produced in cross 1 and one of females produced in cross 2. Cross 3 results in a litter of 12 kits (young rabbits).

Fill in the expected numbers of each of the three types of rabbit in this litter.

•	Agouti rabbits:	
•	Otter rabbits:	١

Black rabbits:

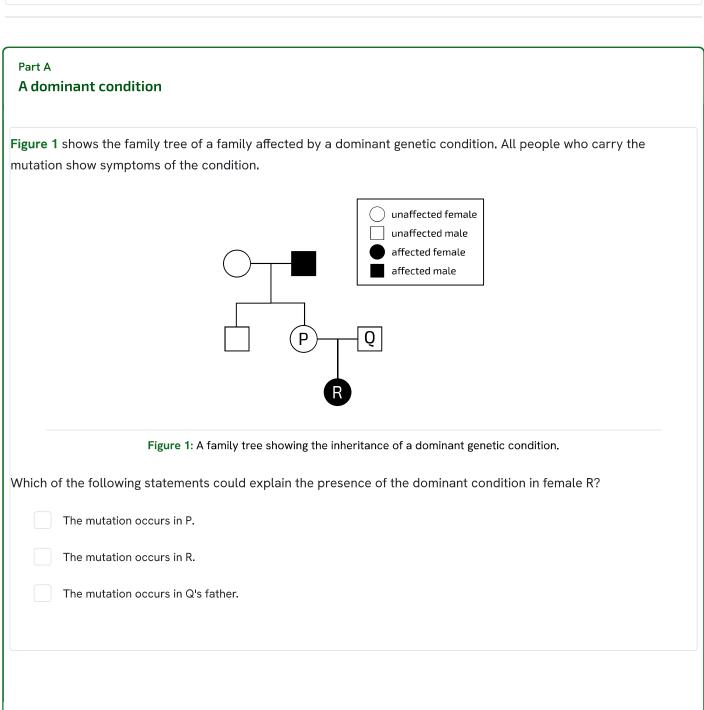
Part D Explanation
In each of the examples above, the dominant allele, A , is thought to be the original allele, and the recessive allele, a , is thought to have been produced by a frameshift mutation in the coding region of the gene.
Which two statements below provide the most likely explanation for the recessive (black hair) phenotype in these three mammals?
The role of the original agouti protein (produced by the dominant allele) is to activate production of eumelanin.
The role of the original agouti protein (produced by the dominant allele) is to inhibit production of eumelanin.
The protein produced by the recessive allele has a slightly different amino acid sequence but performs the same function, just slightly less effectively.
The protein produced by the recessive allele has a slightly different amino acid sequence but performs the same function, just slightly more effectively.
The protein produced by the recessive allele has a completely different amino acid sequence and therefore has a completely different function.
The protein produced by the recessive allele has a completely different amino acid sequence and therefore is non-functional.

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Dominant and Recessive Conditions

Subject & topics Biology Genetics Inheritance		
Status Not started	Stage & difficulty A Level Challenge 2	



Part B

Cystic fibrosis and sickle cell anaemia

Cystic fibrosis and sickle cell anaemia are both recessive genetic conditions and the genes for these conditions are found on different <u>autosomes</u>.

The following statements are true for one set of parents who only have one child:

- Both parents are heterozygous for cystic fibrosis
- One parent is homozygous recessive for sickle cell anaemia
- One parent is heterozygous for sickle cell anaemia

What is the probability of this child having both conditions?

Part C

Pedigree probability

Figure 2 shows the inheritance of a phenotypic feature caused by a recessive allele. "A" represents an individual of unknown genotype.

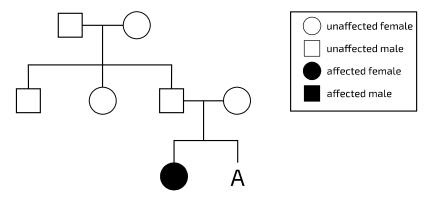


Figure 2: A family tree showing the inheritance of a recessive genetic condition.

What is the probability that individual A is an unaffected male?

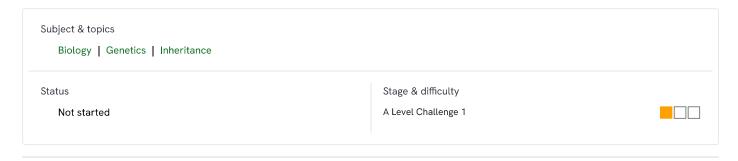
Assume that no mutations occurred to this gene in this family tree.

Adapted with permission from NSAA 2018 Section 1 Q65, NSAA 2021 Section 2 Q41 & NSAA 2021 Section 1 Q74

Question deck:



Eyesight Problems



A gene found on the X chromosome in humans has two alleles: dominant and recessive. Individuals who only have recessive alleles have a condition that affects their eyesight. The inheritance of the condition is shown in **Figure 1**.

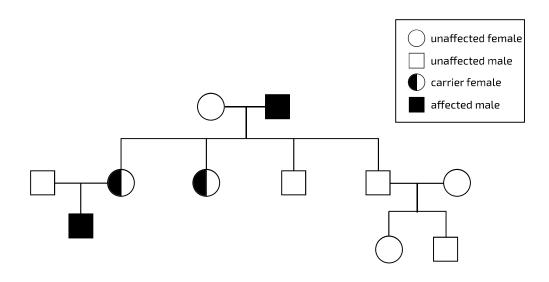


Figure 1: A family tree showing X-linked recessive inheritance. "Unaffected" individuals do not have the condition, "affected" individuals do have the condition, and "carrier" individuals do not have the condition but do carry the recessive allele. The key is shown in the top-right.

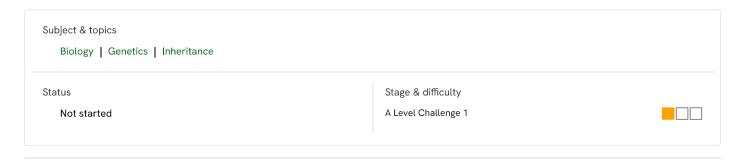
Part A Recessive & dominant alleles
If one living skin cell from each member of this family were analysed, how many recessive alleles (for this gene) would be found?
If one living skin cell from each member of this family were analysed, how many dominant alleles (for this gene) would be found?
Part B An affected female and an unaffected male
If an affected female mates with an unaffected male to produce a son, what is the probability that this individual has the condition?
If an affected female mates with an unaffected male to produce a daughter, what is the probability that this individual
has the condition?

Part C X-linked recessive inheritance
Which of the following statements are true of X-linked recessive inheritance? Select all that apply.
All male offspring of an affected female will have the condition, regardless of the male parent's genotype.
Females cannot have the condition but they can be carriers.
Males are more likely to have the condition than females.
Males cannot be carriers: they either have the condition or they do not.
All female offspring of an affected male will have the condition, regardless of the female parent's genotype.
Females are more likely to have the condition than males.
Adapted with permission from NSAA 2019 Section 1 Q68

Question deck:



Rabbit Fur Colour

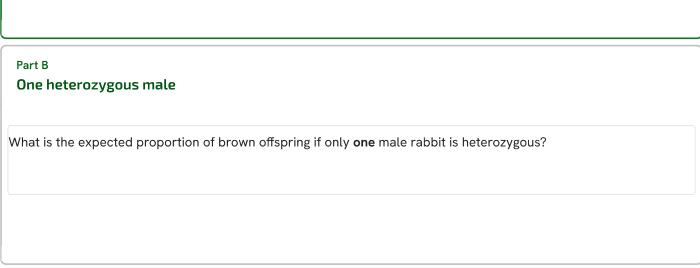


In rabbits, there are two alleles concerned with dark pigment in the fur:

- the dominant allele, B, for black colour
- the recessive allele, b, for brown colour

Two male black rabbits of unknown genotype each mated with a different female brown rabbit.

Part A Two heterozygous males What is the expected proportion of brown offspring if both male rabbits are heterozygous?



Adapted with permission from NSAA 2020 Section 1 Q79

Question deck:



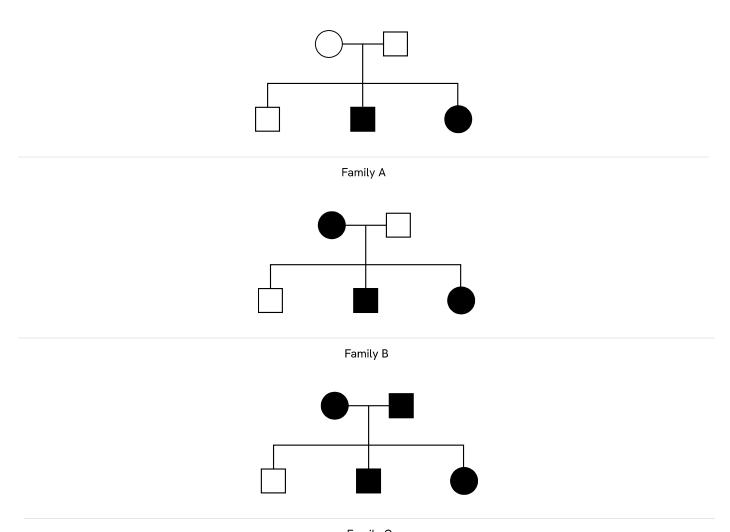
Three Family Trees

Subject & topics Biology Genetics Inheritance		
Status	Stage & difficulty	
Not started	A Level Challenge 1	

The three family trees below show the inheritance of three different genetic conditions, each controlled by one gene with one dominant and one recessive allele.

Circles represent females and squares represent males.

Black circles/squares represent affected individuals and white circles/squares represent unaffected individuals.



Family C

In the questions below, assume that no new mutations occur and that the genes are not found on the X chromosome.

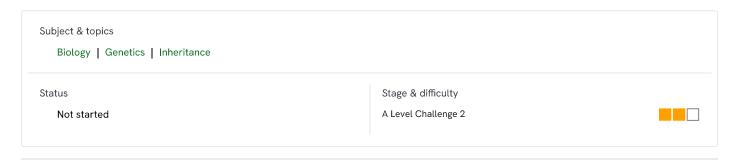
Part A Heterozygous fathers	
In which of the family trees must the male parent be heterozygous? Select all that apply.	
Family A	
Family B	
Family C	
Part B Dominant conditions	
In which of the family trees must the condition be a dominant genetic condition? Select all that apply.	
Family A	
Family B	
Family C	
Part C Recessive conditions	
In which of the family trees must the condition be a recessive genetic condition? Select all that apply.	
Family A	
Family B	
Family C	

Adapted with permission from NSAA 2020 Section 1 Q80

Question deck:



Blood Types



Part A

The ABO gene

The ABO gene codes for an enzyme involved in modifying cell-surface glycoproteins. Once modified, these glycoproteins act as antigens (cell-surface molecules that the immune system can recognise as self or non-self). There are multiple alleles for this gene, and these can be split into three main types: I^A, I^B, and i (alternatively named I^O). The I^A allele produces type A antigens and the I^B allele produces type B antigens. The i allele produces a non-functional enzyme, and therefore does not produce type A or type B antigens.

The table below shows how these alleles relate to blood type.

Blood Type	Antigens produced	Genotype
AB	type A and type B	I _A I _B
А	type A only	I ^A I ^A or I ^A i
В	type B only	I ^B I ^B or I ^B i
0	neither	ii

Fill in the blanks below.

• Allele I ^A is	allele I ^B and	allele i.
• Allele IB is	allele I^A and	allele i.
• Allele i is	allele I^A and	allele I^B.

Items:

dominant to	recessive to	codominant with

Part B The Rh genes
The Rh genes are a group of genes that encode red blood cell membrane transporters. One of these genes, the RhD gene, encodes a membrane transporter that can act as
allele, one i allele, one RhD positive allele, and one RhD negative allele, would have the blood type
Items: dominant O+ O- A+ codominant an antigen an antibody recessive A-
Part C
Blood types and blood transfusion
Individuals will produce antibodies against antigens that their own cells If, during a blood transfusion, an individual receives blood cells of an incompatible type, an immune response will be triggered against these blood cells. Antibodies binding to blood cells can cause the blood cells to clump together, which can cause blood vessel blockage and rupture. O- individuals antibodies against A antigens, B antigens, and RhD antigens. Therefore, they can receive
during a blood transfusion. Their blood cells can be given to any other blood type, because they lack antigens. Because of this, O- individuals are called "universal donors".
AB+ individuals antibodies against A antigens, B antigens, and RhD antigens. Therefore, they can receive during a blood transfusion. Because of this, AB+ individuals are called "universal recipients".
Items:
produce do not produce only O- blood cells blood cells of any type only AB+ blood cells

Part D Offspring blood type(s)
In one family, the following blood types are present:
Mother: A+Father: O-
Which of the following blood types could their children have? Select all that apply.
Assume no mutations occur.
AB+
AB-
A+
A-
B+
B-
O+
O-

Part E Maternal blood type(s)
In another family, the following blood types are present:
• Father: AB-
Child 1: A- Child 2: B-
Which of the following blood types could the mother have? Select all that apply.
Assume no mutations occur.
AB+
AB-
A+
A-
B+
B-
O+
o-

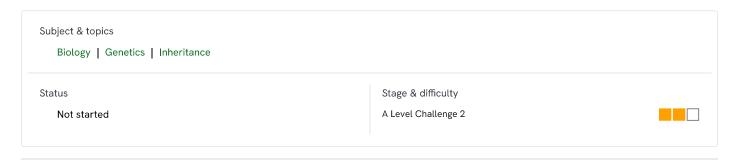
Part F Paternal blood type(s)
In another family, the following blood types are present:
Mother: O-
Child 1: B- C
Child 2: A+
Which of the following blood types could the father have? Select all that apply.
Assume no mutations occur.
AB+
AB-
A+
A
B+
B-
O+
0-

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Question deck:



Squash Plant Genetics



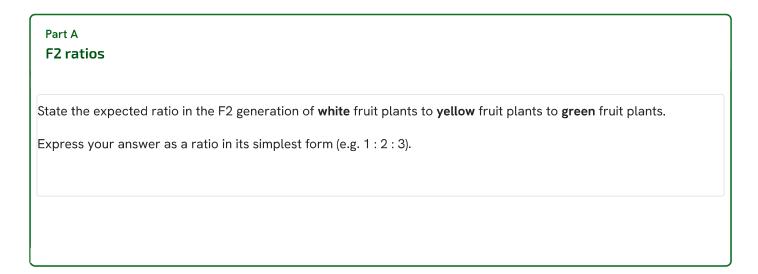
The patty pan squash plant, Cucurbita pepo, produces edible fruits that vary in colour.

The colour of the fruits is controlled by two genes, A/a and B/b, that occur on different chromosomes.

- Allele A produces a white colour.
- Allele **a** does not produce a colour by itself but allows the colours coded by gene **B/b** to show in the phenotype.
- Allele **B** produces a yellow fruit colour.
- Allele **b** produces a green fruit colour.

In a dihybrid cross, an AABB plant was crossed with an aabb plant. All the resulting F1 plants produced white fruits.

The F1 plants were then crossed with each other to obtain the F2 generation.



Part B

Test crosses

Test crosses were carried out on two white-fruited plants, **P** and **Q**, from the F2 generation. Each of these plants had its female flowers pollinated with pollen from a green-fruited plant.

For plant \mathbf{P} , half of the offspring were white and half were yellow.

For plant \mathbf{Q} , half of the offspring were white and half were green.

State the genotype of plant P .
State the genotype of plant Q .

Part C

Genetic variation

Plants P and Q show genetic variation with respect to fruit colour alleles.

Name the process that occurred during meiosis in the F1 parents that produced this variation.

During which phase of meiosis does this process occur?

Adapted with permission from CIE A Level Biology, June 2020, Paper 4, Question 2a