

GENETICS

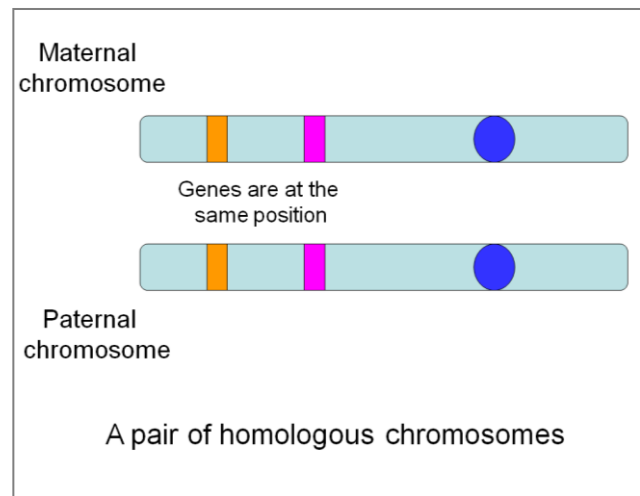
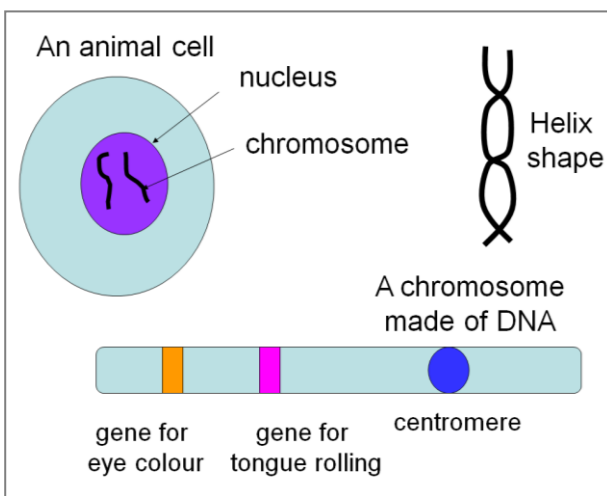
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NAME: _____

DNA AND GENETICS RECAP AND DEFINITIONS

- Our body is made up of _____
- Each cell contains a _____
- _____ are found inside the nucleus
- Each chromosome is made up of a long molecule of _____
- This molecule is twisted into a _____ shape

-
- A _____ is a length of DNA that codes for a _____
 - Each gene is responsible for a _____
 - E.g. there is a gene that codes for eye colour and another that codes for tongue rolling



- there are **46** _____ of every cell in the body
- chromosomes occur in pairs called _____
- Chromosomes are passed from one generation to the next _____
- _____ chromosome pair comes from the male **sperm** cell and the other from the female **ovum**
- Therefore we have some characteristics like our mother and some like our father

-
- homologous chromosomes _____
 - _____
 - Genes may have different forms called _____
 - E.g. the gene for eye colour has the alleles blue and brown

- The alleles which you have is called your _____
- The characteristics which they produce is called your _____ (think "ph" **physical** features/characteristics - the **phenotype**)
- Some alleles are _____ over others and if they are present in the genotype they will always be _____
- Other alleles are _____ and you must have _____ for the characteristic to be expressed in the phenotype
- Every cell has _____
- If each allele is the same the cell is _____ for that gene
- Homozygous alleles can be _____
- If the alleles are different they are _____

Read through this example carefully:



EXAMPLE

- The allele for brown eyes, B is dominant to the allele for blue eyes b.
- A person with genotype BB is homozygous and their phenotype will be brown eyes
- A person with genotype Bb is heterozygous and their phenotype will be brown eyes
- A person with genotype bb is homozygous and their phenotype will be blue eyes

REMEMBER

- _____ (pollen, eggs, sperm and ova) are made by a special form of cell division and contain _____
- Sex cells are called _____ because they contain half a set of chromosomes
- All other cells (hair, skin, liver etc) are called _____ because they contain 2 of each chromosome

Genetics in pea plants

Gregor Mendel's monohybrid cross:

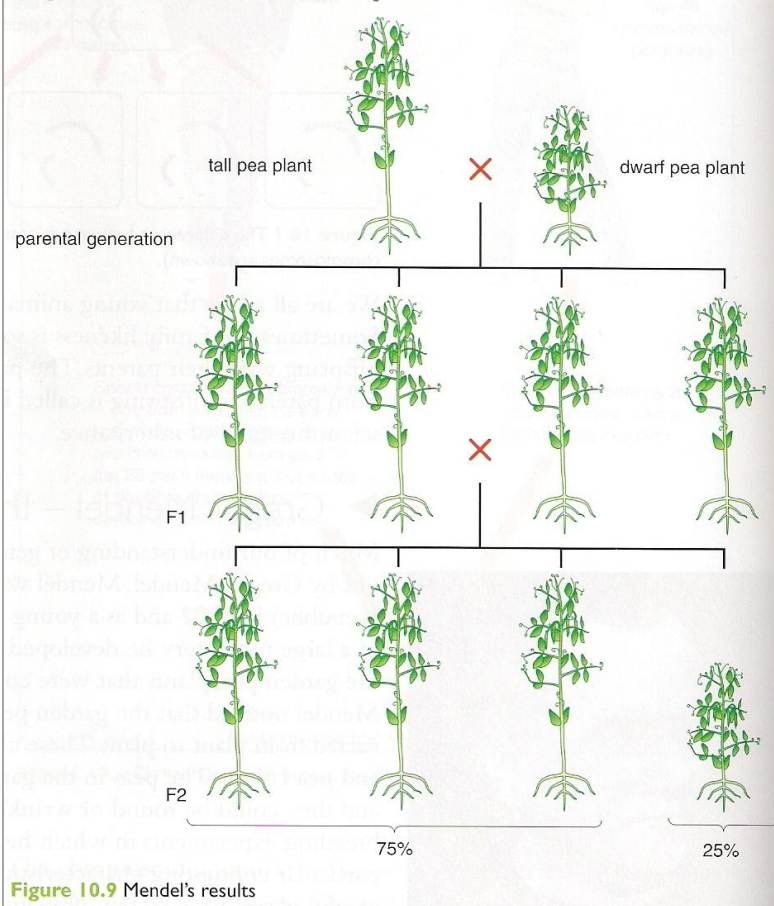
Mendel noticed that pea plants (like all organisms) _____
e.g. different pea shape and colour, and height of plant



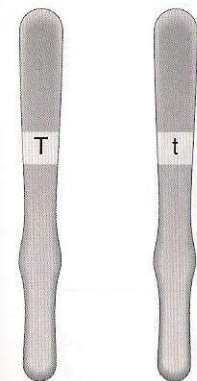
He _____ (mated) them through _____
to study how a single characteristic (e.g. height of plant) was passed on to the next generation (_____) i.e. _____

We now know that these characteristics were caused by the genes (alleles) on the _____ being passed on - depending on which _____ the offspring received from the parents

Explanation of the monohybrid cross



The position of one gene on a pair of chromosomes:



In this example the two alleles of the gene are different. The individual is heterozygous for the characteristic concerned

Usually one allele will be _____ over the other and in order for the recessive allele to be seen in the _____ (physical appearance) of the organism, it must be present on _____ chromosomes of the offspring i.e. be _____

Before selecting his parent plants he often let them breed until all offspring produced showed the same characteristic - these parent plants were then called _____ for that characteristic

During meiosis it is totally random which chromosome (either the one originally from the mother, or the one originally from the father) enters which of the 4 daughter cells/gametes. _____

Pea plant example and how we represent the alleles of a cross:

The alleles of a gene are represented by the same letter

..... allele = letter

..... allele = case

e.g. height in pea plants

.....

.....

As all cells contain pairs of homologous chromosomes a pea plant can have the following genotypes and phenotypes:

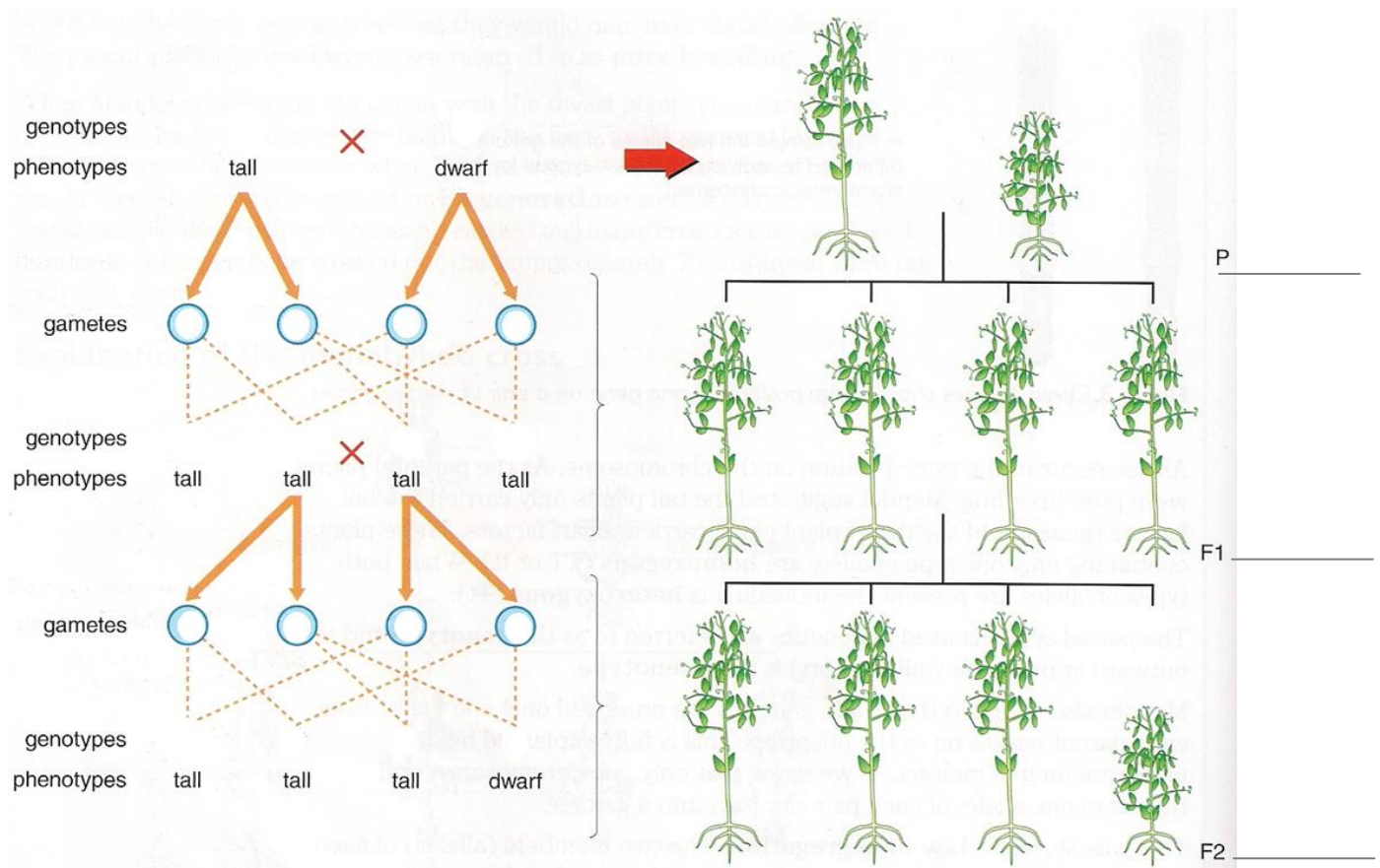
Genotype	Phenotype

Pollen and ovules produced during meiosis only have one of each allele, so:

- A pea plant with genotype can only produce pollen or eggs with the allele
- A pea plant with genotype can produce pollen or eggs with the allele or allele
- A pea plant with genotype can only produce pollen or eggs with the allele

Complete the diagram to show Mendel's monohybrid cross for the characteristic of plant height. _____

REMEMBER the law of segregation: Only one allele will enter each gamete:



The inheritance of most human characteristics is complex, usually involving many genes but some features like eye colour and the ability to roll your tongue show monohybrid inheritance i.e. a single allele causes the characteristic. We can predict how such characteristics are passed on from parents to offspring using genetic diagrams.

When completing these monohybrid crosses for certain characteristics e.g. what Mendel did with height in pea plants, it is helpful to use a _____

In these the potential gametes/alleles from each parent's genotype are crossed to see what the potential outcomes are for the offspring (i.e. their genotypes and phenotypes)

E.g. In the following example we see the inheritance of eye colour in humans (B = brown, b = blue)



father's gametes

	mother's gametes	
	B	B
b	Bb	Bb
b	Bb	Bb



100% of offspring have brown eyes
100% of offspring are heterozygous Bb



father's gametes

	mother's gametes	
	B	b
b	Bb	bb
b	Bb	bb



50% of the offspring are heterozygous Bb and will be expected to have brown eyes

50% are homozygous bb and will be expected to have blue eyes



father's gametes

	mother's gametes	
	B	b
B	BB	Bb
b	Bb	bb



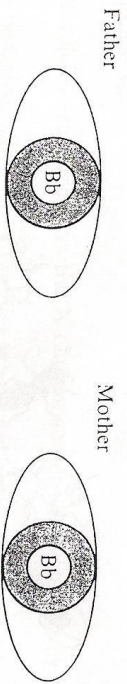
So there is a 75% chance of these two heterozygous brown-eyed parents having a brown-eyed child, and a 25% chance of having a child with blue eyes. This gives the ratio 3:1 brown:blue. This percentage chance applies at each conception, therefore it is possible for all the children to have blue eyes.

READ THESE GENETICS RULES CAREFULLY TO HELP YOU PREDICT THE OUTCOME OF CROSSES!

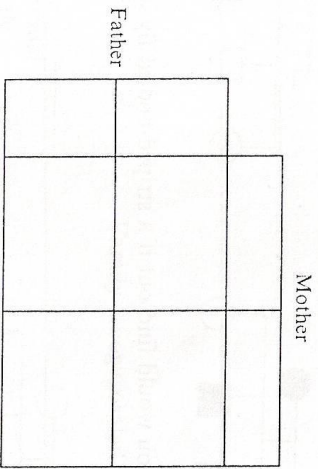
- Ratios will only be accurate when large numbers of offspring are produced. This is because it is totally random which gametes, and therefore alleles fuse during fertilisation
- If both parents are homozygous for the same allele the offspring will be too
- If one parent is homozygous dominant and the other heterozygous then all offspring will show the dominant phenotype, though half will be heterozygous in genotype
- If one parent is homozygous dominant and the other homozygous recessive, all offspring will be heterozygous with the dominant phenotype
- If one parent is heterozygous and the other homozygous recessive, half the offspring will be heterozygous with the dominant phenotype and the other half will be homozygous recessive with the recessive phenotype i.e. 1:1 phenotype ratio
- If both parents are heterozygous, 25% of the offspring will be homozygous dominant, 50% will be heterozygous and 25% will be homozygous recessive. This gives a phenotype ratio of 75% dominant to 25% recessive i.e. 3:1 phenotype ratio

1

(c) The diagram shows a cross between two people with brown eyes. The allele for brown eyes (B) is dominant to the allele for blue eyes (b).



(i) Complete the Punnett square to show this cross.



(ii) Draw a circle round the homozygous recessive child. [1]

(iii) What is the ratio of brown eyes to blue eyes in the children? [1]

Ratio _____ [1]

(iv) Name the structures on which the alleles are found. [1]

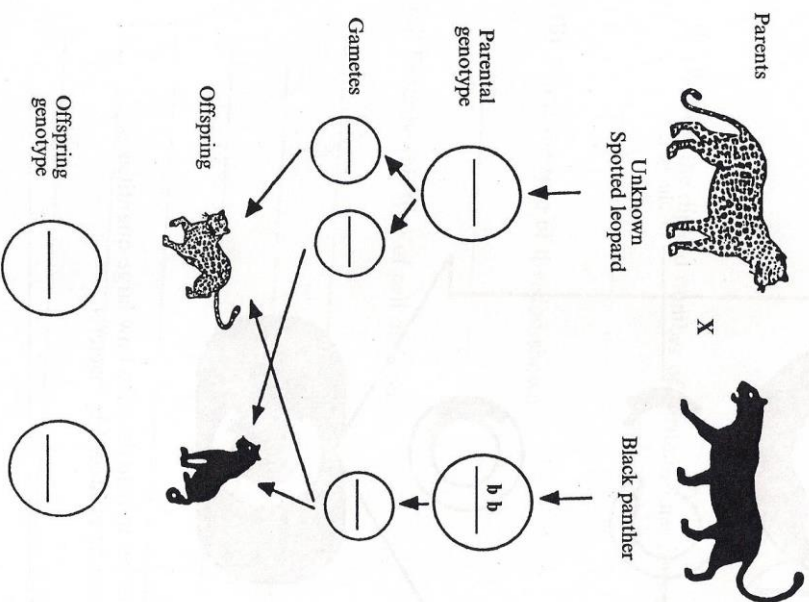
_____ [1]

2

Leopards and panthers belong to the same species. Their coat colour is controlled by a pair of alleles.

Let **B** represent the dominant allele for spotted coat, and **b** represent the recessive allele for black coat.

(a) Complete the diagram by writing **BB**, **Bb**, **bb**, **B** or **b** in each of the circles provided.



[5]

(b) Why is the genotype of a black panther always known? [1]

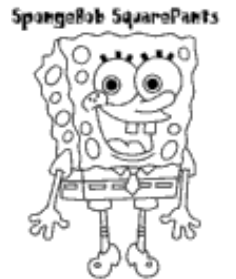
_____ [1]

Spongebob Genetics

Scientists at Bikini Bottom have been investigating the genetic makeup of the organisms in this community. Use the information provided and your knowledge of genetics to answer each question.

1. For each genotype below, indicate whether it is a heterozygous (He) OR homozygous (Ho).

TT _____ Bb _____ DD _____ Ff _____ tt _____ dd _____
 Dd _____ ff _____ Tt _____ bb _____ BB _____ FF _____



2. Determine the phenotype for each genotype using the information provided about SpongeBob.

Yellow body colour is dominant to blue.

YY _____ Yy _____ yy _____

Square shape is dominant to round.

SS _____ Ss _____ ss _____

3. For each phenotype, give the genotypes that are possible for Patrick.

A tall head (T) is dominant to short (t).

Tall = _____ Short = _____

Pink body colour (P) is dominant to yellow (p).

Pink body = _____ Yellow body = _____

4. SpongeBob SquarePants recently met SpongeSusie Roundpants at a dance. SpongeBob is heterozygous for his square shape, but SpongeSusie is round. Create a Punnett square to show the possibilities that would result if SpongeBob and SpongeSusie had children. HINT: Read question 2!

A. List the possible genotypes and phenotypes for their children.

B. What are the chances of a child with a square shape? _____

C. What are the chances of a child with a round shape? _____

5. Patrick met Patti at the dance. Both of them are heterozygous for their pink body colour, which is dominant over a yellow body colour. Create a Punnett square to show the possibilities that would result if Patrick and Patti had children. HINT: Read question 3!



A. List the possible genotypes and phenotypes for their children.

B. What are the chances of a child with a pink body? _____

C. What are the chances of a child with a yellow body? _____

6. Everyone in Squidward's family has light blue skin, which is the dominant trait for body colour in his hometown of Squid Valley. His family brags that they are a "purebred" line. He recently married a nice girl who has light green skin, which is a recessive trait.



Create a Punnett square to show the possibilities that would result if Squidward and his new bride had children. Use B to represent the dominant gene and b to represent the recessive gene.

A. List the possible genotypes and phenotypes for their children.

B. What are the chances of a child with light blue skin? _____

C. What are the chances of a child with light green skin? _____

D. Would Squidward's children still be considered purebreds? Explain!

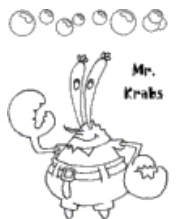
7. Assume that one of Squidward's sons, who is heterozygous for the light blue body colour, married a girl that was also heterozygous. Create a Punnett square to show the possibilities that would result if they had children.

A. List the possible genotypes and phenotypes for their children.

B. What are the chances of a child with light blue skin? _____

C. What are the chances of a child with light green skin? _____

8. Mr. Krabbs and his wife recently had a Lil' Krabby, but it has not been a happy occasion for them. Mrs. Krabbs has been upset since she first saw her new baby who had short eyeballs. She claims that the hospital goofed and mixed up her baby with someone else's baby. Mr. Krabbs is homozygous for his tall eyeballs, while his wife is heterozygous for her tall eyeballs. Create a Punnett square using T for the dominant gene and t for the recessive one.



A. List the possible genotypes and phenotypes for their children.

B. Did the hospital make a mistake? Explain your answer.

THE TEST CROSS (BACK CROSS)

Sometimes in agriculture or in breeding domestic animals, it is important to know the genotype of a particular animal or plant with the dominant phenotype i.e. whether it is _____.
The _____ can reveal this.

e.g. you want to buy a Dalmatian dog, breed it and sell the puppies. BUT, a friend has told you that some Dalmatians have brown spots, not black and that these dogs are sometimes difficult to sell.

This happens because there are 2 alleles for spot colour:

- dominant B=black,
- recessive b=brown

If two black spot dogs heterozygous for spot colour are crossed, some puppies may be brown (remember it is random which gametes from the parents actually fuse but we can still look at probable ratios of offspring).



So how can you tell if your dog showing the dominant phenotype is homozygous or heterozygous for spot colour?

How it works:

The dominant individual with the unknown genotype is _____. If
_____ offspring are produced _____
then the unknown genotype can be discovered by looking at the _____.

- If any of the offspring show the recessive phenotype then the mystery parent genotype must be _____
- If all offspring show the dominant phenotype e.g. black spots, then the individual's genotype was _____ for the dominant allele

To summarise:

To see if your dog is heterozygous or homozygous for black spots you would cross your black spotted dog with a brown spotted dog.
If any of the puppies are brown you know you have a dog heterozygous for spot colour and could end up with brown spot puppies

Have a look at the following test cross then try the questions:

In the example of the pea, a tall plant could be homozygous (TT) or heterozygous (Tt). To identify the unknown genotype of the plant it is crossed with a homozygous recessive plant.

If the unknown genotype is TT

parental phenotype tall × dwarf
genotype TT tt

gametes (T) (t)

Punnett square

	t
T	Tt

offspring [genotype all Tt
phenotype all tall

If the unknown genotype is Tt

parental phenotype tall × dwarf
genotype Tt tt

gametes (T) (t) (t)

Punnett square

	t
T	Tt
t	tt

offspring [genotype 50% Tt 50% tt
phenotype 50% tall
50% dwarf

So if any dwarf plants are produced the unknown parent was heterozygous (Tt).

Try these examples:

1(a). The allele for black hair (H) is dominant to the allele for fair (h). A woman with black hair marries a man with fair hair (homozygous recessive). What colour of hair would their children have? (Draw the 2 possible Punnett squares and list the phenotypic ratios of the offspring)



(b). Explain how this information can be used to determine the mother's genotype: _____

2. A mouse with brown fur is bred with a mouse with white fur (bb). All of the offspring were brown. What is the genotype of the brown mouse? (show how this can be displayed using the back cross)

3. One characteristic of a fly used in genetic experiments is wing length. The allele (gene) for long wings (D) is dominant to the allele (gene) for short wings (d). Frank had a specimen tube containing two flies, one with the genotype DD and the other with genotype Dd. For an experiment he needed the fly with the genotype DD.

(a). Explain why it was not possible for him to identify the genotypes of the two flies just by looking at them: _____

He decided to do two backcrosses (testcrosses) to determine the genotypes of the flies:

(b). Give the genotype of a third fly that he would have to use in each backcross: _____

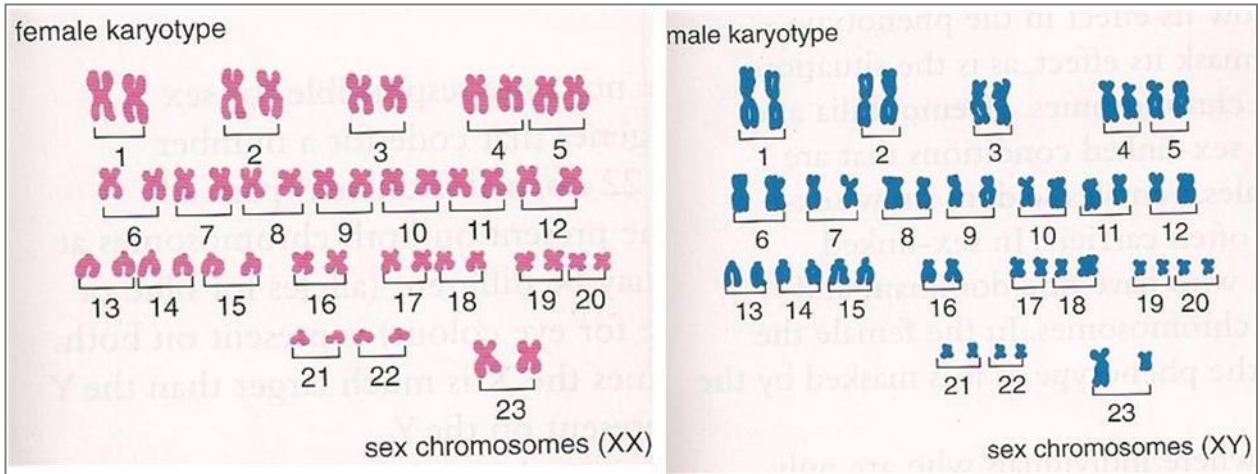
(c). Explain how the results of the two backcrosses would enable him to work out the genotype of each fly: _____



SEX DETERMINATION IN HUMANS

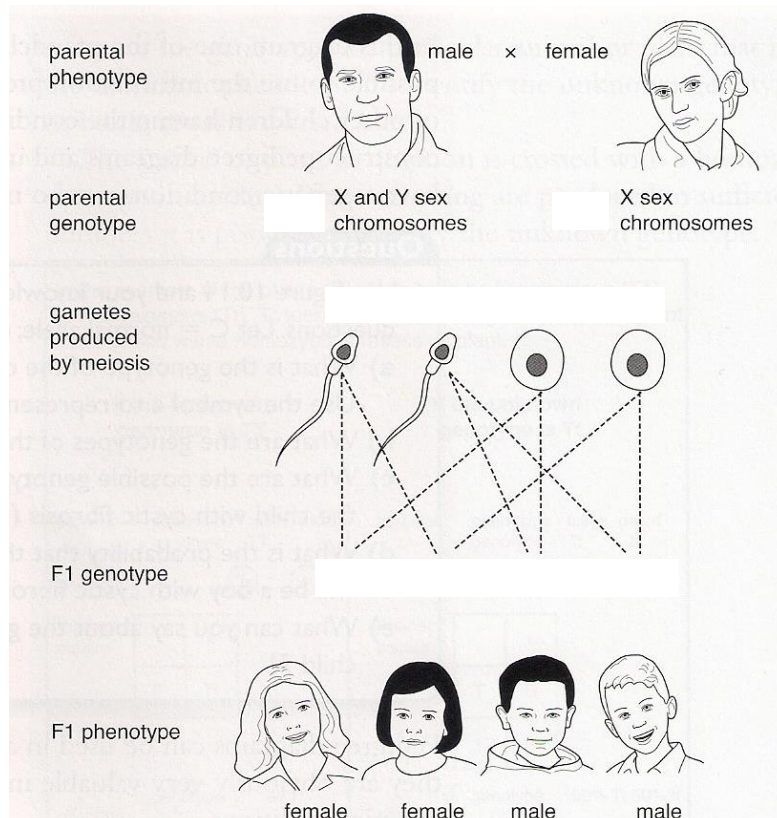


Sex is genetically determined. In a normal human cell (except gametes) there are 22 pairs of normal chromosomes and _____ . These sex chromosomes determine the sex of the individual. _____ have one ___ and one ___ chromosome whereas _____ have _____ chromosomes. A complete set of chromosomes is called a _____. *The complete set of human chromosomes:*



During meiosis the female will provide one X chromosome for each _____ (egg), but half of the male's sperm will have an X chromosome and half will have a Y chromosome. Therefore there is an _____ of an X or Y chromosome from the male being involved in fertilisation and joining with the ovum from the female (containing an X chromosome). Equal numbers of males and females should thus be produced through sexual reproduction.

Complete the diagram to show the sex chromosomes:



REMEMBER: Fertilisation is random! i.e. which sperm fertilises the ovum. We all know of large families consisting of only sons or daughters i.e. not the equal proportion of males and females as expected.

QUESTION: What is the chance/probability of parents having five children, all of whom are male?

- Every time she has a baby the chance of it being a boy is ____ (50% or $\frac{1}{2}$)
- To find the chance of her having 5 boys with her first 5 babies we

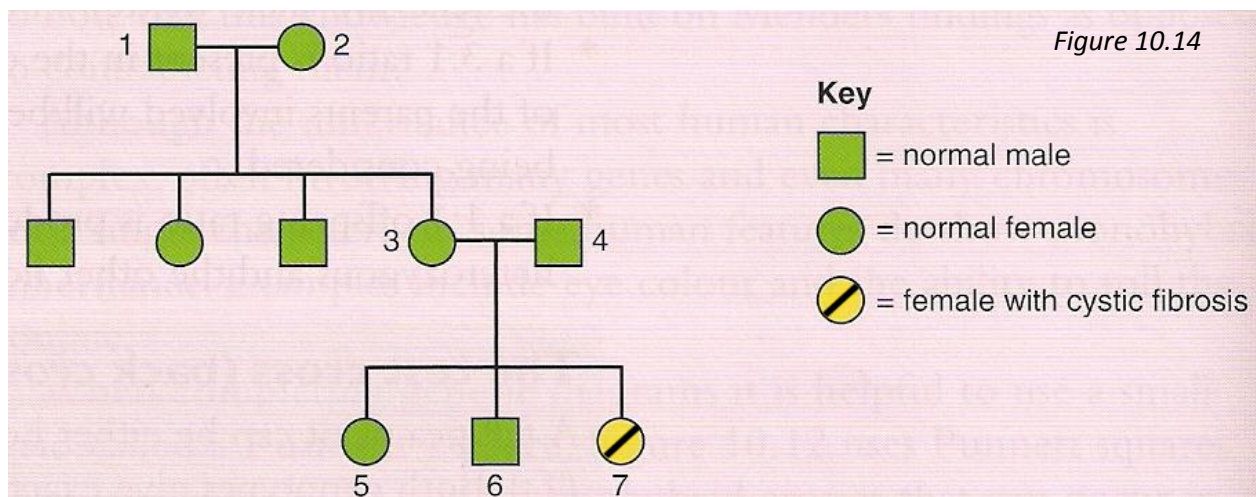
_____:

- _____
- I.e. ($0.5 \times 0.5 \times 0.5 \times 0.5 \times 0.5 = 0.03125$)

INHERITED/GENETIC DISEASES

Some diseases can be inherited, eg cystic fibrosis which is a medical condition caused by a recessive gene where lungs get congested by excess mucous.

These show the way in which a genetic condition is inherited in a group of biologically related people/family. Below is an example showing how cystic fibrosis is inherited:



How many grandchildren will have cystic fibrosis? _____

Using the information we can work out the _____ of other children having the disease. A _____ may do this with a couple who are sufferers or carriers of a genetic disease. You can use pedigree diagrams in any type of genetic cross to follow a characteristic, but they are most useful in tracing and predicting harmful genetic conditions

The X and Y chromosomes are not only responsible for sex determination. They also have genes that code for a number of body functions. Each of the 22 normal (non-sex) pairs of chromosomes has the same gene present on both chromosomes at the same position. The alleles may be different (alleles for blue or brown eyes) but the gene (gene for eye colour) is present on both. However, in the sex chromosomes the X is much larger than the Y and carries genes that are not present on the Y.

This is particularly important in males as they only have one X chromosome. Therefore any recessive allele carried on an X chromosome in a male will show its effect in the phenotype – there is no dominant allele to mask its effect, as is the situation with females who have two X chromosomes. Haemophilia and red-green colour blindness are sex-linked conditions that are almost exclusively found in males. Females seldom show sex-linked conditions but they are often carriers.

- What is different about the sex chromosomes compared to the normal chromosome pairs? _____

- Why is this particularly important for males? _____

- Why will a recessive allele carried on the X chromosome in a male show its affect in the phenotype? _____

- Although females rarely show sex linked conditions like red-green colour blindness and haemophilia, what role do they often play? _____

These carriers don't show the phenotype of the disease because they have one dominant allele on one X chromosome and one recessive allele on the other. In the female the recessive allele does not affect the phenotype as it is masked by the dominant allele.

Haemophilia is a condition where individuals who are only carrying the recessive allele are unable to make all the products required to _____ the blood. Individuals with red-green colour blindness are _____

The inheritance of haemophilia: Complete your diagrams

H = normal allele h = haemophiliac allele

Cross 1 haemophiliac male × normal female

parental genotype

gametes ○○ ○

Punnett square

offspring

[genotype	50%	<input type="text"/>	50%	<input type="text"/>
	phenotype	50%	carrier females	50%	normal males

Cross 2 normal male × carrier female

parental genotype

gametes ○○ ○○

Punnett square

offspring

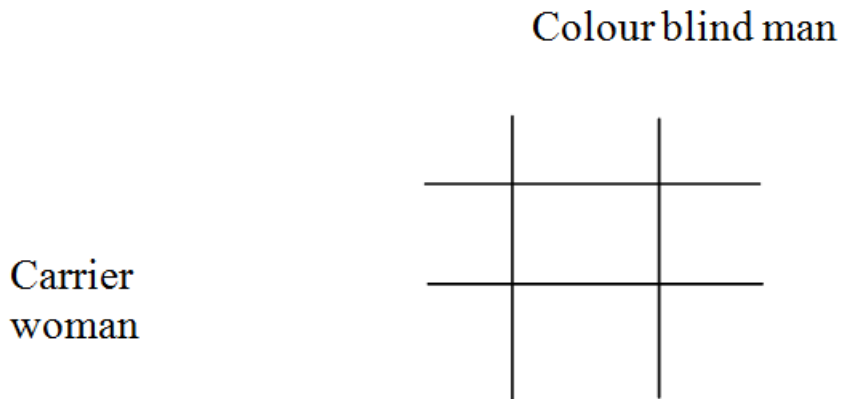
[genotype	25%	<input type="text"/>	25%	<input type="text"/>	25%	<input type="text"/>	25%	<input type="text"/>
	phenotype	25%	normal females	25%	carrier females	25%	normal males	25%	haemophiliac males

These crosses show why haemophilia is usually found only in males. Very occasionally, females may inherit the condition.

QUESTION:

1. a) Certain types of colour blindness are sex-linked. What does this mean?

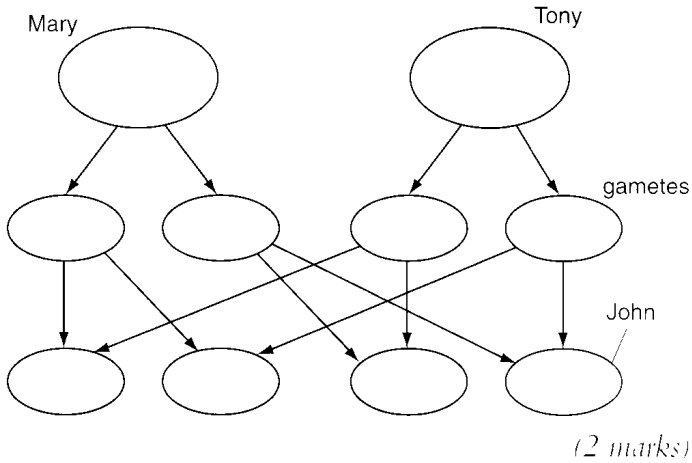
b) Using the symbol X^B to represent the allele for normal colour vision and X^b for colour blindness, draw a diagram to show the possible genotypes and phenotypes of children produced by a colour-blind man and a carrier woman.



Phenotypes: _____

Exam style questions:

- 6 a) Brown eye colour is dominant to blue eyes. Mary has blue eyes and her partner Tony has brown eyes. Their son John has blue eyes. Copy and complete the diagram below to show how John inherits blue eyes. Use symbols: B = brown allele b = blue allele.

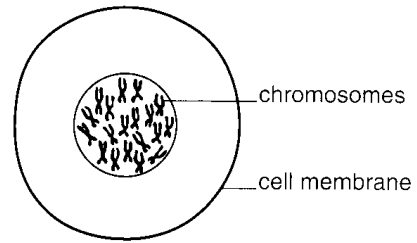


- b) What is the probability (chance) of Mary and Tony's next child having brown eyes? (1 mark)
- c) Using eye colour as an example, explain the term **heterozygous**. (2 marks)
- d) Use your knowledge of sex determination in humans to describe why equal numbers of boys and girls are born. (3 marks)

- 7 Pea plants can produce peas that are wrinkled or smooth. The gene (allele) for wrinkled is dominant to the gene (allele) for smoothness. Let R represent the gene (allele) for wrinkled peas. Let r represent the gene (allele) for smooth peas.

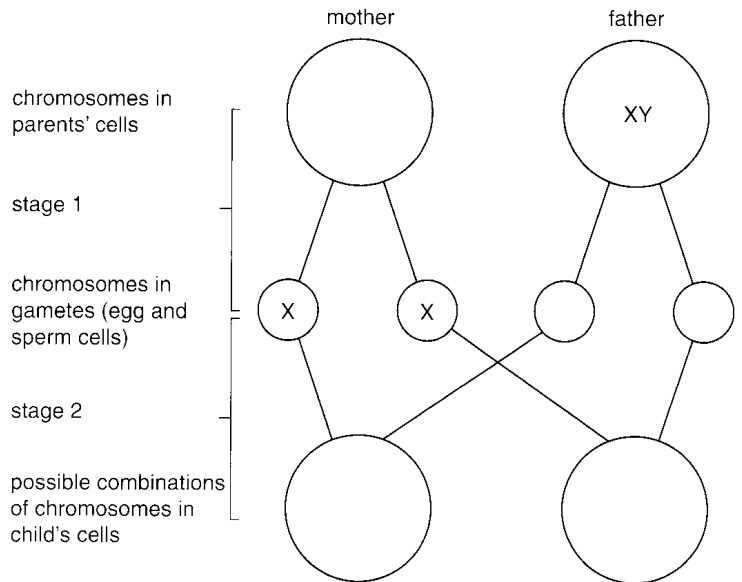
- a) A plant breeder crossed two heterozygous pea plants.
- i) Use a Punnett square to show the possible genotypes of the offspring of this cross. (3 marks)
- ii) Give the phenotypes of the offspring and the ratio of the phenotypes. (2 marks)

- b) The diagram shows a cell with chromosomes.



- i) Where in the cell are the chromosomes located? (1 mark)
- ii) What are short sections of chromosomes called? (1 mark)
- iii) What chemical are chromosomes made from? (1 mark)

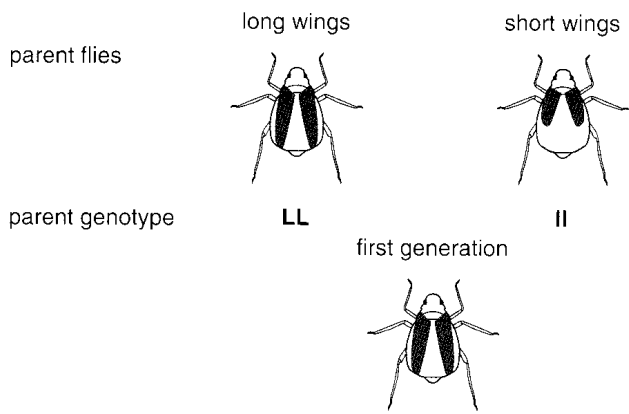
- c) The following diagram shows how the sex of a child depends on the chromosomes it inherits from its parents.



Copy and complete the diagram above to show the sex chromosomes found in each cell.

(3 marks)

8 a) The diagrams show the results of a breeding experiment with fruit flies.

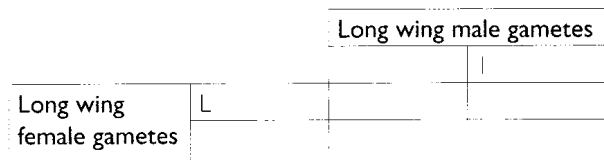


the allele for long wings is L
the allele for short wings is l

- i) Which allele is dominant? (1 mark)
- ii) Give the genotype of the first generation fly. (1 mark)

b) A male and a female from the first generation were crossed.

Copy and complete the Punnett square to show the genotypes of the second generation flies.

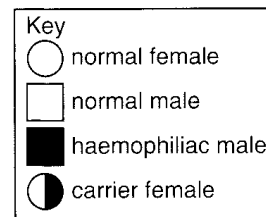
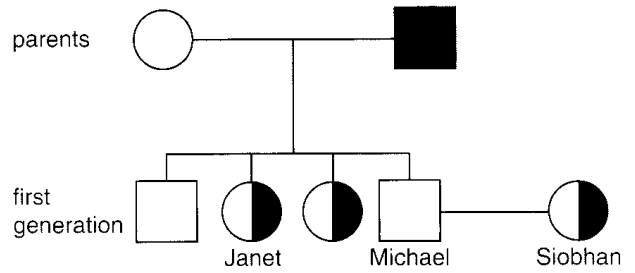


(2 marks)

- c) Geneticists wanted to determine the **genotype** of a long wing fly. Describe the procedure they would have to follow to determine its genotype. **In this question you will be assessed on using your written communication skills including the use of specialist science terms.** (6 marks)

9 Haemophilia is a genetic disorder which is sex-linked.

The diagram shows a family tree.



Let X^H represent a normal X chromosome
 X^h represent an X chromosome carrying the haemophiliac allele
 Y represent a Y chromosome.

- a) Give the genotypes of the parents. (2 marks)
 - b) Give the phenotype and genotype of Janet. (2 marks)
 - c) Explain how Michael is a normal male even though his father is a haemophiliac. (2 marks)
- Michael and Siobhan have children.
- d) What proportion of the children will have haemophilia? (1 mark)
 - e) Why are there fewer haemophiliac females than males? (1 mark)

