GENETICS

2.3.12-14

Genetic Diagrams and Terminology	2.3.12	understand and interpret genetic diagrams consisting of a single characteristic controlled by a single gene with two alleles (monohybrid cross) in plants, animals and humans (*w – all of (iii)*): – dominant and recessive alleles; – genotype, phenotype, gamete and offspring ratios, percentages and probabilities; – homozygous and heterozygous genotypes; – Punnett squares to determine genotype frequencies; – ***test (back) crosses to determine an unknown genotype;*** and – ***pedigree diagrams*** (*w – all of (iii)*);

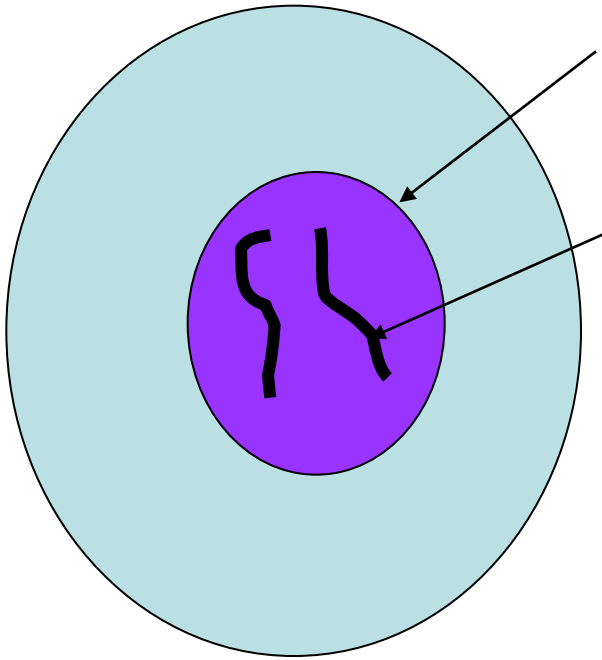
Watch "Genetic inheritance" video clip

(0:00-~3:20)

DNA and genetics recap and definitions...

- Our body is made up of **cells**
- Each cell contains a **nucleus**
- **Chromosomes** are found inside the nucleus
- Each chromosome is made up of a long molecule of **DNA**
- This molecule is twisted into a **helix** shape
- A **gene** is a length of DNA that codes for a **protein**
- Each gene is responsible for a **different characteristic**
- E.g. there is a gene that codes for eye colour and another that codes for tongue rolling

An animal cell



nucleus

chromosome

Helix shape

A chromosome made of DNA

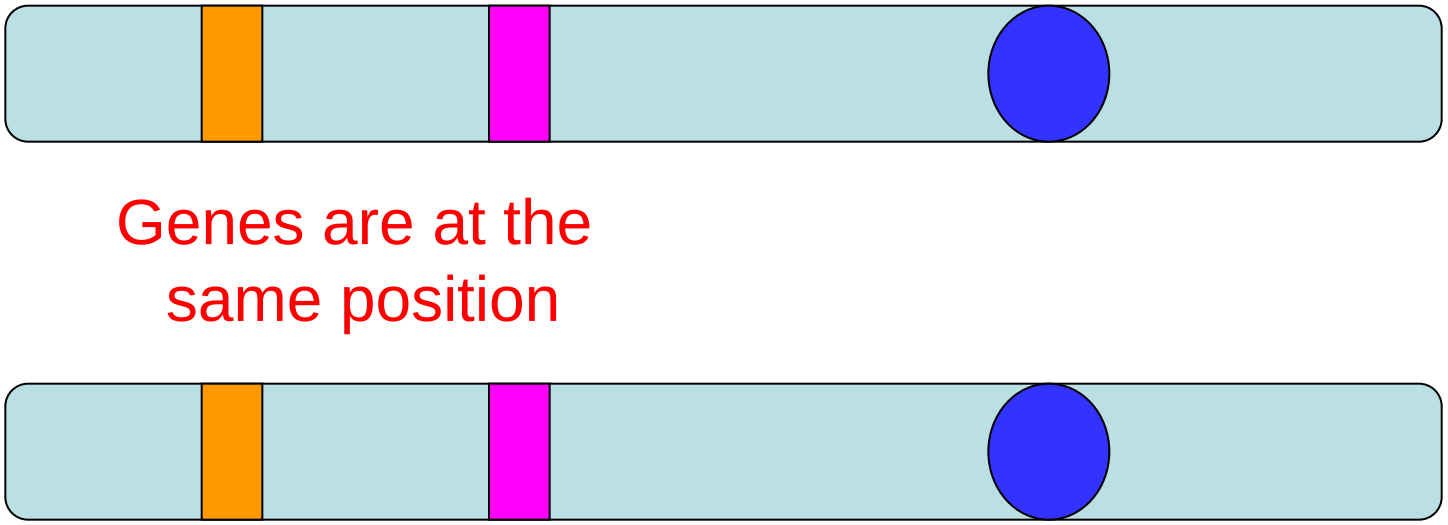
centromere

gene for eye colour

gene for tongue rolling

- There are **46 chromosomes (23 pairs)** in the nuclei of every cell in the body (apart from the sex cells)
- chromosomes occur in pairs called **homologous chromosomes**
- Chromosomes are passed from one generation to the next **at fertilisation**
- **One of each** 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

Maternal chromosome



Genes are at the same position

Paternal chromosome

A pair of homologous chromosomes

- Homologous chromosomes **carry the same genes at the same position**
- Genes may have different forms called **alleles**
- E.g. the gene for eye colour has the alleles blue and brown
- The alleles which you have is called your **genotype**
- The characteristics which they produce is called your **phenotype** (think "ph" **ph**ysical features/characteristics – the **ph**enotype)
- Some alleles are **dominant** over others and if they are present in the genotype they will always be **expressed in the phenotype**
- Other alleles are **recessive** and you must have **2 copies of these alleles** for the characteristic to be expressed in the phenotype

- Every cell has 2 copies of each gene (one on each chromosome in a pair)
- If each allele is the same the cell is homozygous for that gene
- Homozygous alleles can be dominant or recessive e.g. BB or bb
- If the alleles are different they are heterozygous e.g. Bb

EXAMPLE

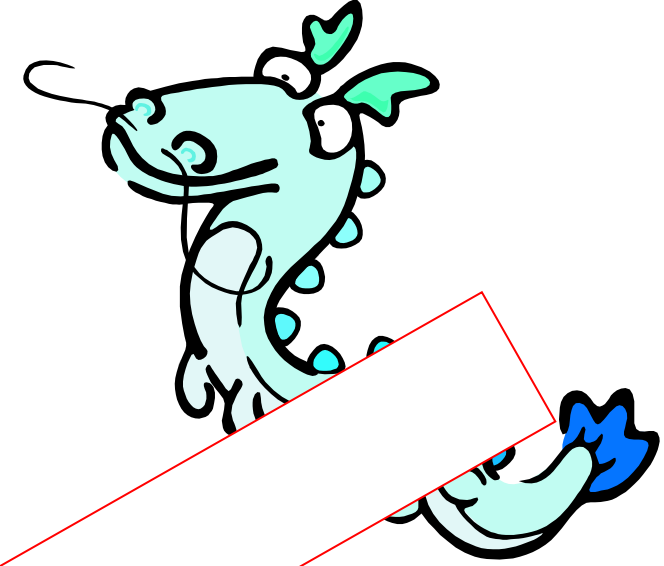
The allele for brown eyes, B is dominant to the allele for blue eyes b.

- 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

- **Sex cells** (pollen, eggs, sperm and ova) are made by a special form of cell division and contain **only one chromosome** from each pair.
- Sex cells are called **HAPLOID CELLS** because they contain half a set of chromosomes
- All other cells (hair, skin, liver etc) are called **DIPLOID CELLS** because they contain 2 of each chromosome.

Dragon genetics



Optional

What does this teach u

- **The dominance of** **over recessive ones**
- **The genetic** **n produced by sexual reprodu** **e fertilisation of gametes and the pairi** **andomly segregated chromosomes from parents creates genetically different offspring**

Instructions

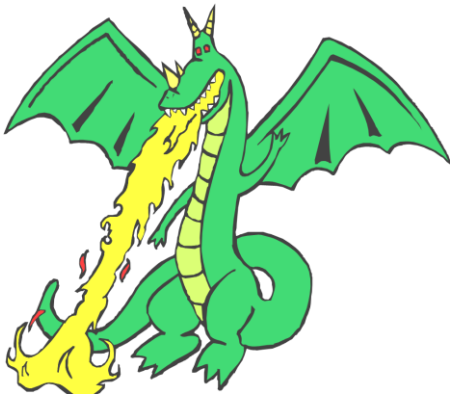
1. Throw 2 sticks of the same colour
2. Record the letters on the sticks
3. Use the key to find out the characteristic for your dragon baby
4. Repeat with the other sticks
5. Draw your dragon baby

Red stick	W-wings	w-no wings
Green stick	A-purple skin	a-green skin
Yellow stick	R-red eyes	r-black eyes
Blue stick	S-back spines	s-no spines
Purple stick	T- 4 toes	t-3 toes
Pink stick	P-spots	p-no spots
Brown stick	B-long tail	b-short tail
Black Stick	F-fire breather	f-non fire breather
Grey stick	E-round eyes	e-oval eyes
Orange stick	N-nose spike	n-no nose spike
L. Blue Stick	G-tail spikes	g-no tail spikes
White stick	L-short arms	l-long arms

Our Dragon Baby

Colour of Stick	Gene 1	Gene 2	Feature

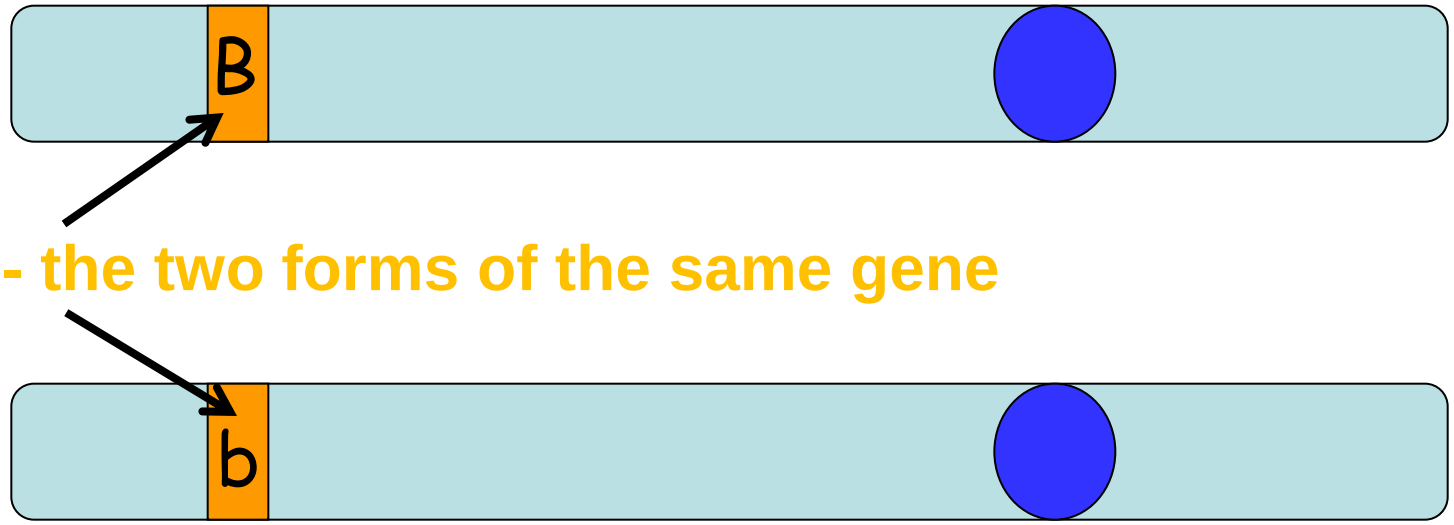
Picture of the Baby



understand and interpret genetic diagrams consisting of a single characteristic controlled by a single gene with two alleles (monohybrid cross) in plants, animals and humans (*w – all of (iii)*):

A pair of homologous chromosomes e.g. no. 14

Maternal chromosome



Alleles - the two forms of the same gene

Paternal chromosome

Qu: What is the genotype for this gene? **Heterozygous**

REMEMBER: During meiosis only one of these alleles will go into a gamete. This is random and is known as Mendel's "law of segregation"

www.bbc.co.uk/schools/gcsebitesize/science

Edexcel: classification, inheritance & variation: genes &inheritance

www.zerobio.com/videos/monohybrid.html

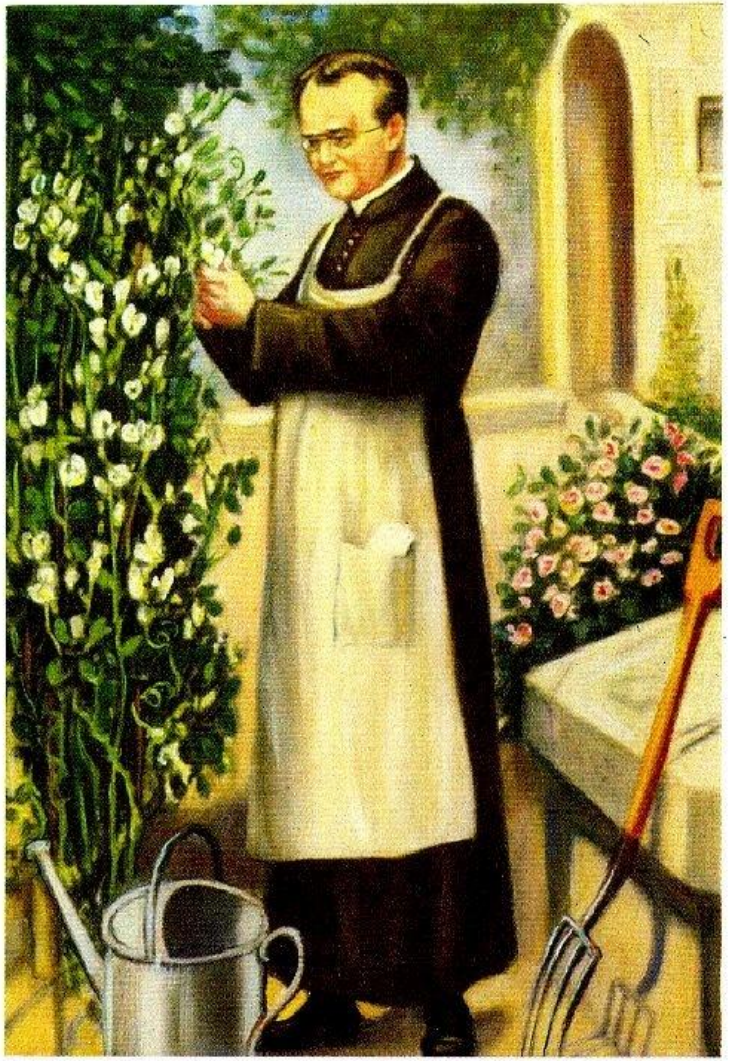
www.sumanasinc.com/webcontent/animations/biology.html

Mendel's experiments

www.kscience.co.uk/animations/anim_1.htm

eye colour

http://www/siskiyous.edu/class/bio1/genetics/monohybrid_v2.html



GREGOR MENDEL!


Pea plants

Mendel's monohybrid cross

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

He **crossed** (mated) them through **cross pollination** to study how a single characteristic (e.g. height of plant) was passed on to the next generation (**F1**) i.e. ***A monohybrid cross***

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

Explanation of the monohybrid cross

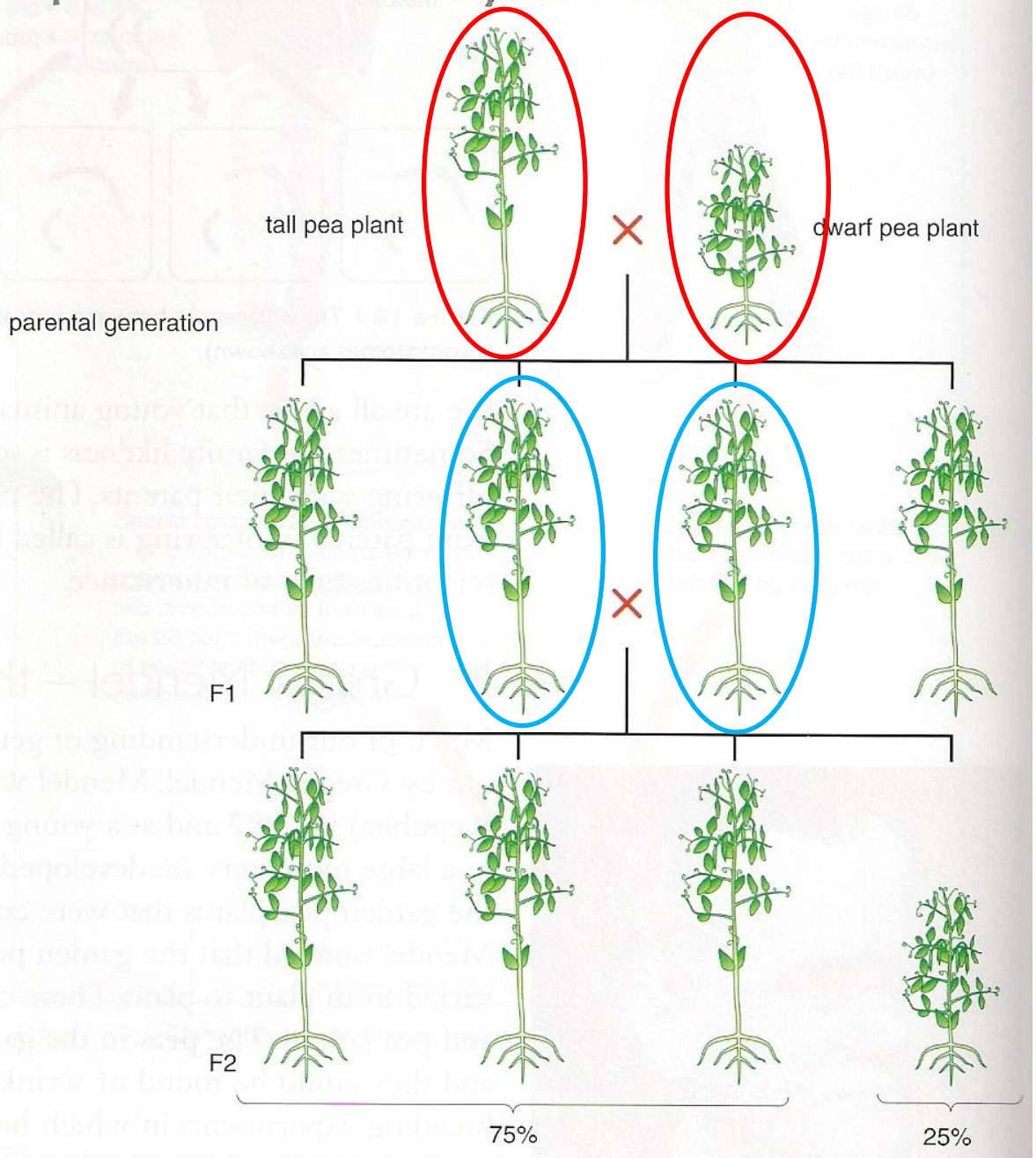
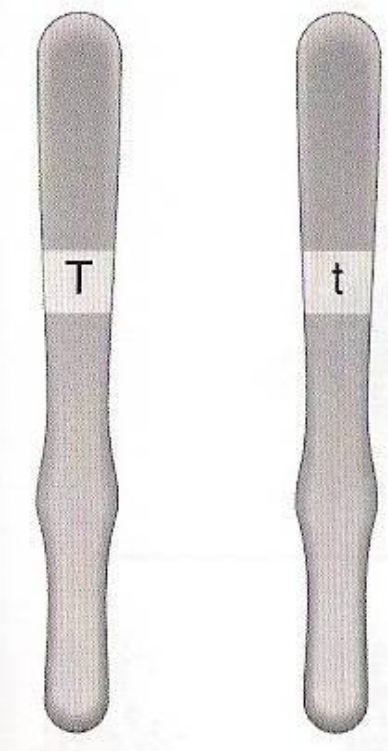


Figure 10.9 Mendel's results



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 **dominant** over the other and in order for the recessive allele to be seen in the **phenotype** (physical appearance) of the organism, it must be present on **both** chromosomes of the offspring i.e. be **homozygous recessive**

Before selecting his parent plants he often let them breed until all offspring produced showed the same characteristic – these parent plants were then called **pure breeding** 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. **This is what Mendel called the "law of segregation" – that only one allele will be present in a gamete**

REMEMBER which gametes fuse at fertilisation to create the new offspring is also random! i.e. all offspring will show genetic variation

11 August 2010 Last updated at 17:59

Pea plant grows inside man's lung

A Massachusetts man who was rushed to hospital with a collapsed lung came home with an unusual diagnosis: a pea plant was growing in his lung.

Can the humble pea grow anywhere?

Ron Sveden had been battling emphysema for months when his condition deteriorated.

He was steeling himself for a cancer diagnosis when X-rays revealed the growth in his lung.

Doctors believe that Mr Sveden ate the pea at some point, but it "went down the wrong way" and sprouted.

"One of the first meals I had in the hospital after the surgery had peas for the vegetable. I laughed to myself and ate them," Mr Sveden told a local Boston TV reporter.

Ron Sveden: "I was kinda surprised"

00.00 / 00.25

Mr Sveden said the plant was about half an inch

- The alleles of a gene are represented by the **same** letter

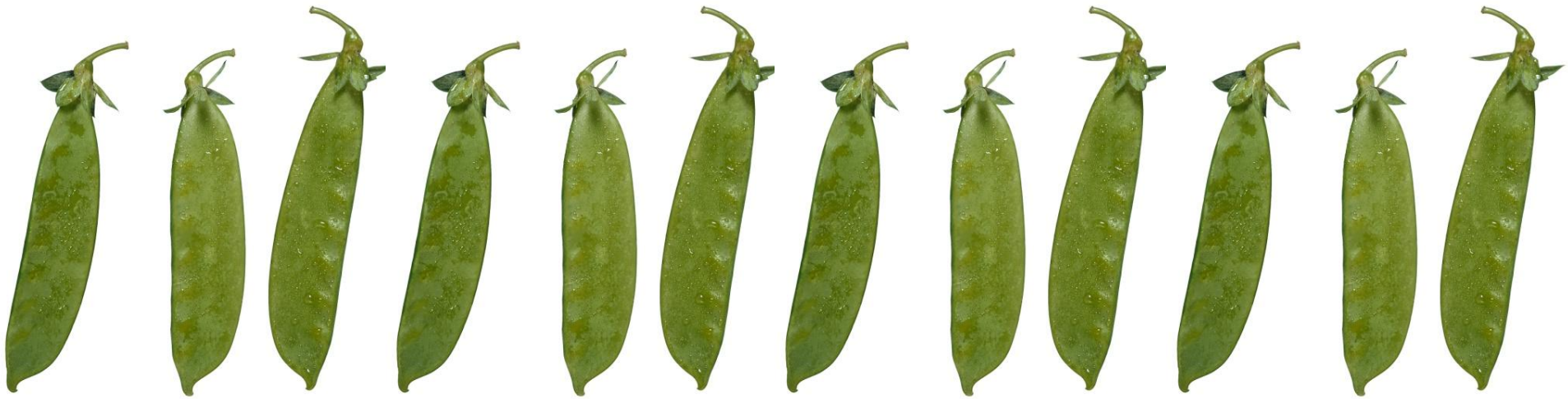
Dominant allele = **capital** letter

Recessive allele = **lower** case

e.g. height in pea plants

T = dominant tall

t = recessive small



- As all cells contain pairs of homologous chromosomes (pairs of alleles), what are the possible genotypes and phenotypes for the height gene in pea plants?

Genotype	Phenotype

- A pea plant with genotype **TT** can only produce pollen or eggs with the allele **T**
- A pea plant with genotype **Tt** can produce pollen or eggs with the allele **T** or allele **t**
- A pea plant with genotype **tt** can only produce pollen or eggs with the allele **t**

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

This shows all the possible ways the gametes of each parent can fuse and the resulting genotypes of the offspring

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

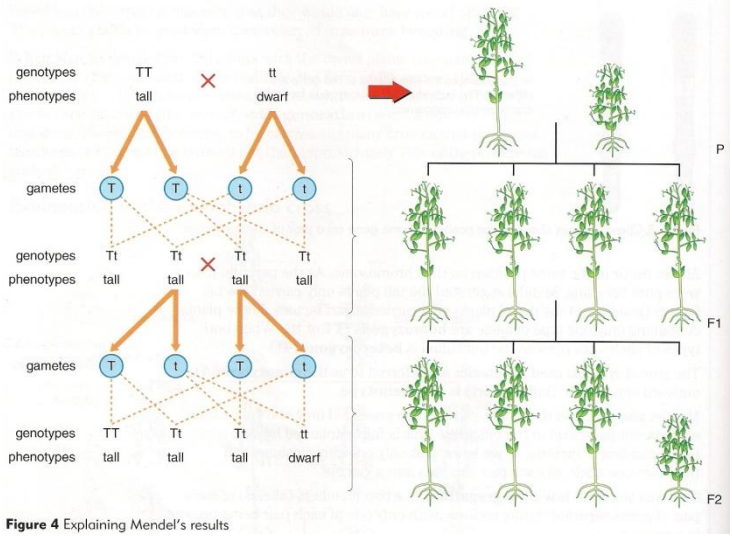


Figure 4 Explaining Mendel's results

Complete the diagram:

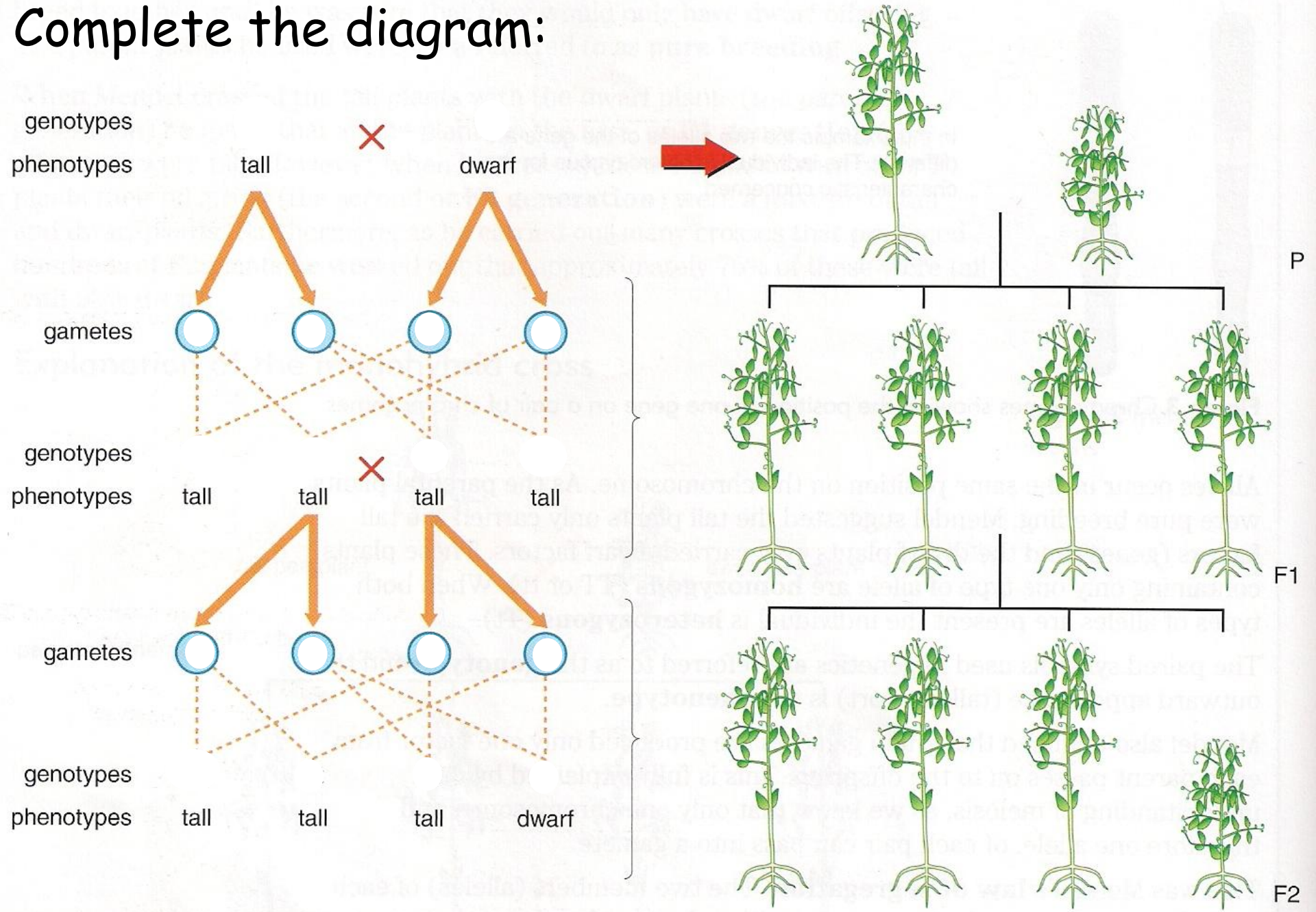


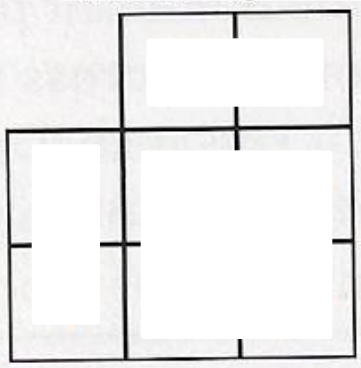
Figure 4 Explaining Mendel's results

- 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 **Punnett square/Genetic test cross diagram.** In these the potential gametes/alleles from each parent are crossed to see what the potential outcomes are for the offspring...

Blue eyed father *(Homozygous recessive)*

Brown eyed mother *(Homozygous dominant)*



Potential offspring genotypes

The inheritance of eye colour in humans (B = brown, b = blue)

Blue eyed father *(Homozygous recessive)*

Brown eyed mother *(Heterozygous)*

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

Brown eyed father *(Heterozygous)*

Brown eyed mother *(Heterozygous)*

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!

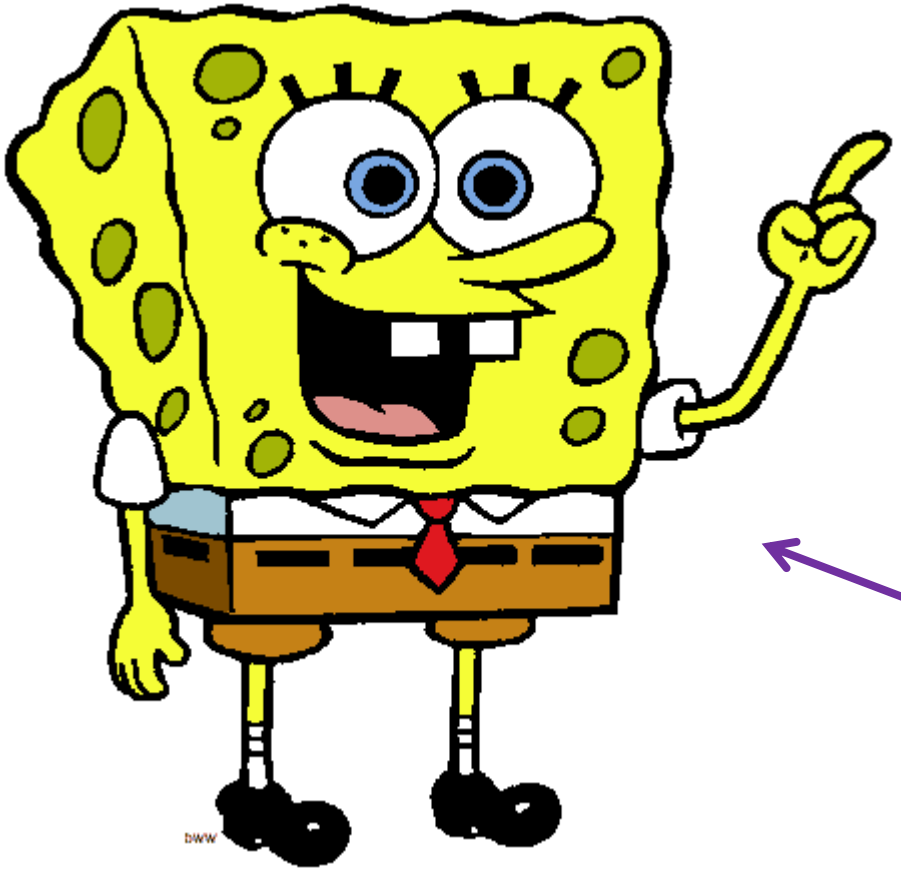
1. Ratios will only be accurate when large numbers of offspring are produced. This is because it is totally random which gametes, and therefore alleles fuse during fertilisation
2. If both parents are homozygous for the same allele the offspring will be too
3. If one parent is homozygous dominant and the other heterozygous then all offspring will show the dominant phenotype, though half will be heterozygous in genotype

Genetics Rules Continued

4. If one parent is homozygous dominant and the other homozygous recessive, all off spring will be heterozygous with the dominant phenotype
5. 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
6. 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

COMPLETE THE 2 PAST PAPER PUNNETT SQUARE QUESTIONS...

TRY MY GENETICS CROSS QUESTIONS FOR HOMEWORK



Spongebob Punnettsquare pants!

Harry Potter and the Recessive Allele

How Are Wizards Made?

Optional for more crosses practice – RM Shared – Mr Dorman (also at end of power point)

Optional for more genetic cross/Punnett square practice - RM Shared – Mr Dorman

monster genetics

Questions 1-7

monster genetics

Questions 8-10

ACTIVITY

BEAKER 1 = 20 brown beads

BEAKER 2 = 20 blue beads

Blindfolded pupil selects one bead from each beaker

Why would all offspring be brown eyed?

Repeat using:

BEAKER 1 = 10 brown & 10 blue beads

BEAKER 2 = 20 blue beads

Calculate the phenotype ratio of brown to blue eyed offspring

Collate class results

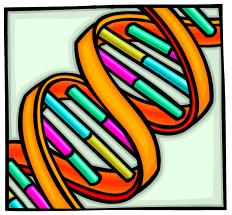
The Test Cross (Back Cross)

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** _____ **or** _____ The **test cross** 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. These dogs are difficult to sell

This happens because there are 2 alleles for spot colour:

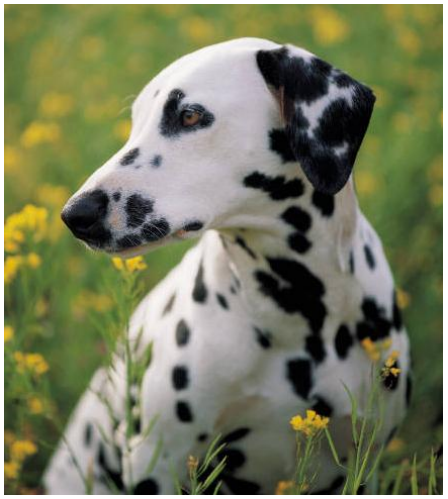
- dominant B = black,
- recessive b = brown



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)

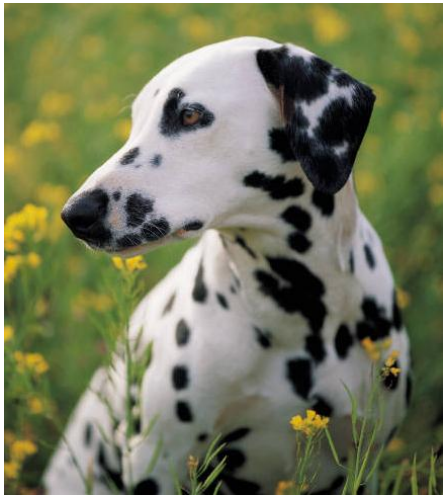
So how can you tell if your dog showing the dominant phenotype is homozygous or heterozygous for spot colour?

Perform a test cross (back cross)



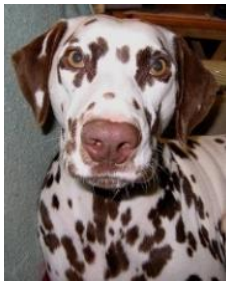
BB

Or?



Bb

How it works:

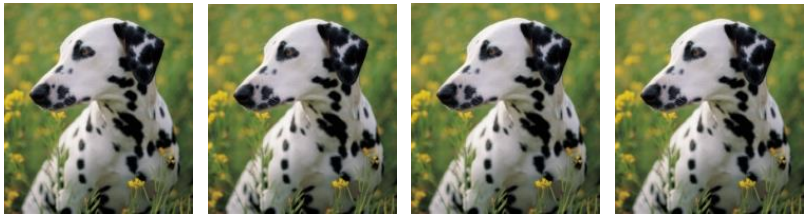


- The dominant individual with the unknown genotype is **crossed with a homozygous recessive individual**:
- If offspring are produced **in sufficient numbers** then the unknown genotype can be discovered by looking at the **phenotype ratios of the offspring**

- If any of the offspring show the recessive phenotype then the mystery parent genotype must be **heterozygous**

Bb

- If all offspring show the dominant phenotype e.g. black spots, then the individual's genotype was **homozygous** for the dominant allele

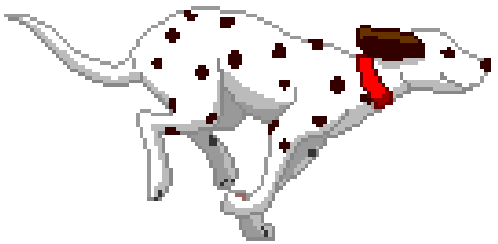


BB

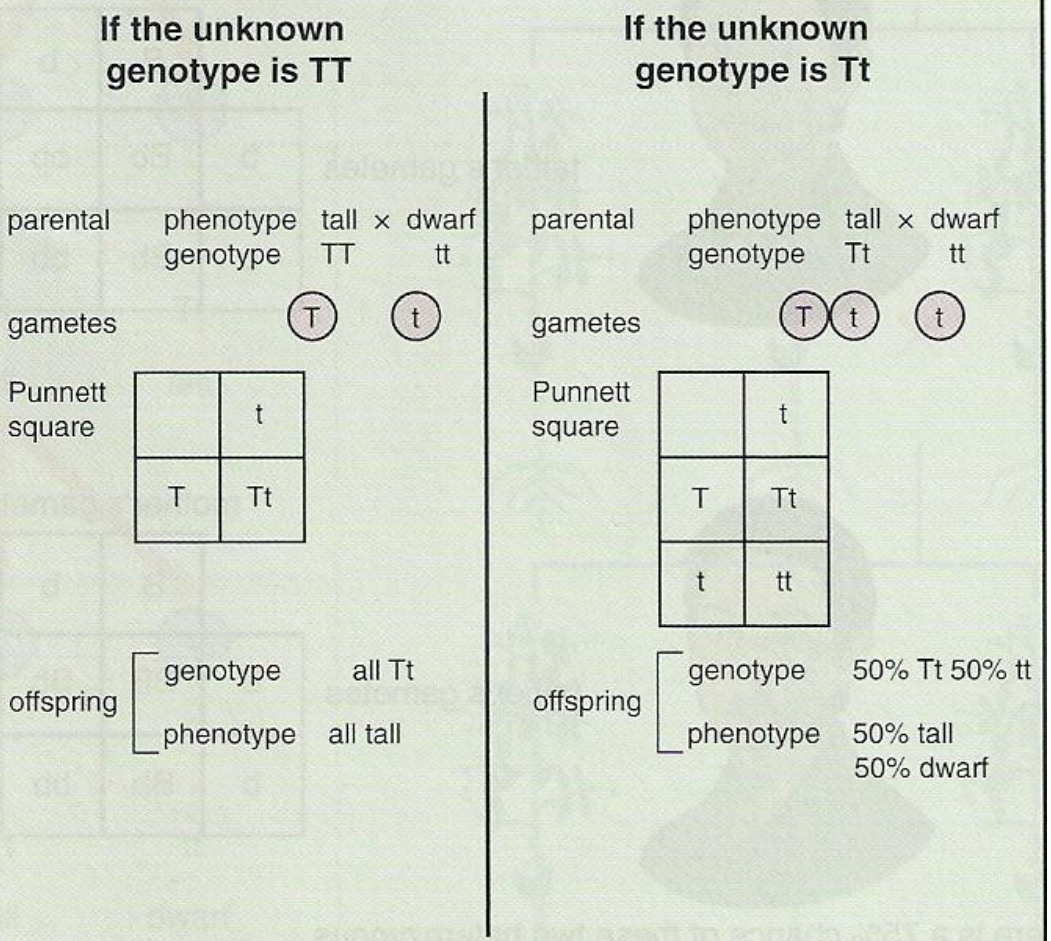
To summarise:

To see of 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



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.



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

Figure 10.13 The test cross

Try the test cross questions...

pedigree diagrams

The X and Y Chromosomes	2.3.13	understand how sex is determined in humans; and
	2.3.14	***understand and explain how some genetic disorders are sex linked (the inheritance of haemophilia and red/green colour blindness)*** *(w – all of (iii))*.

sex

determination

Worksheet

gender

GENDER ACTIVITY

BEAKER 1 = 20 black pipe cleaners (female X chromosome)

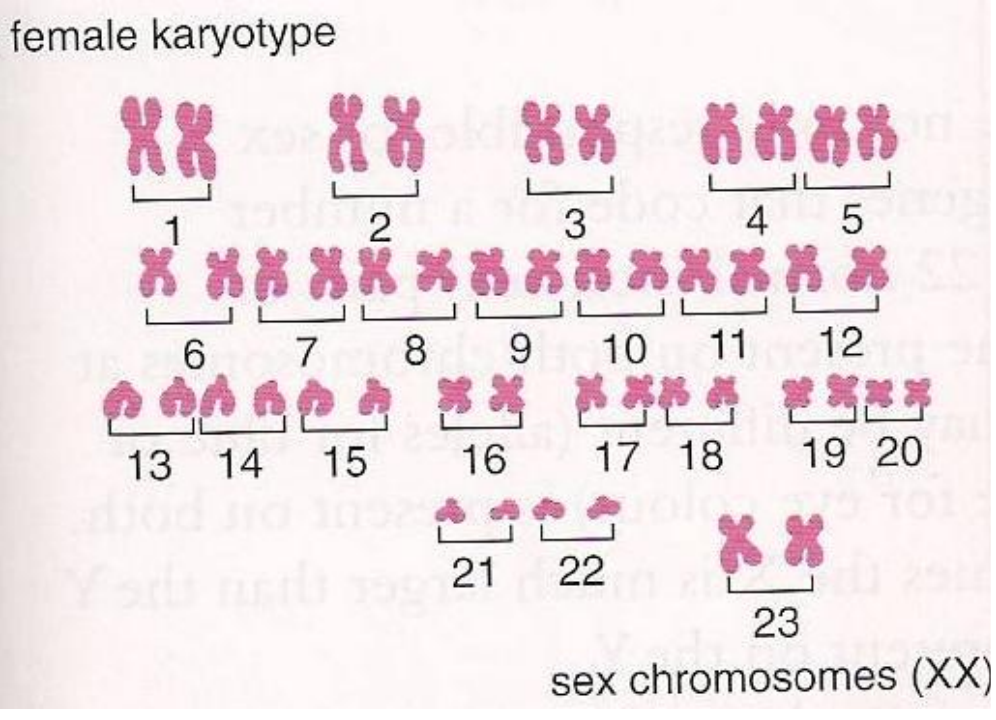
BEAKER 2 = 10 black pipe cleaners (male X chromosome)

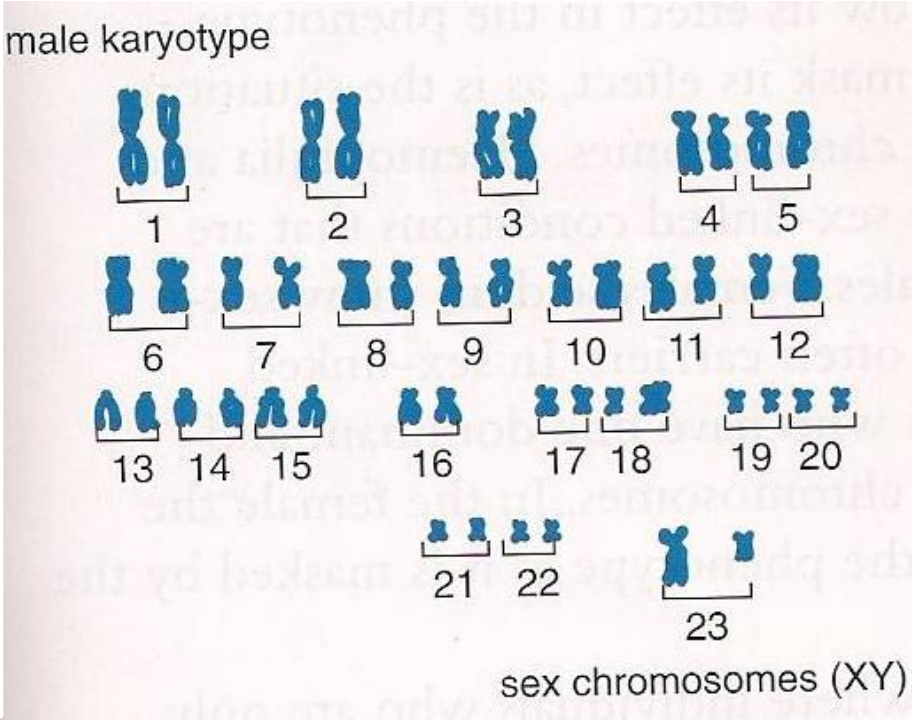
& 10 white pipe cleaners (male Y chromosome)

- Blindfolded pupil selects one pipe cleaner from each beaker
- Record the genotypes & phenotypes
- Calculate the ratio of XX (females) to XY (males)
- Use a Punnet square to explain outcomes.

Sex is genetically determined. In a normal human cell (except gametes) there are 22 pairs of normal chromosomes and **one pair of sex chromosomes**. These sex chromosomes determine the sex of the individual. **Males** have one **X** and one **Y** chromosome whereas **females** have **two X** chromosomes. A complete set of chromosomes is called a **karyotype**.

The complete set of human chromosomes:





During meiosis the female will provide one X chromosome for each **ovum** (egg), but half of the male's sperm will have an X chromosome and half will have a Y chromosome. **Therefore there is an equal chance 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.

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?

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 X 0.5 X 0.5 X 0.5 X 0.5 = 0.03125)

Complete your diagram to show the sex chromosomes:

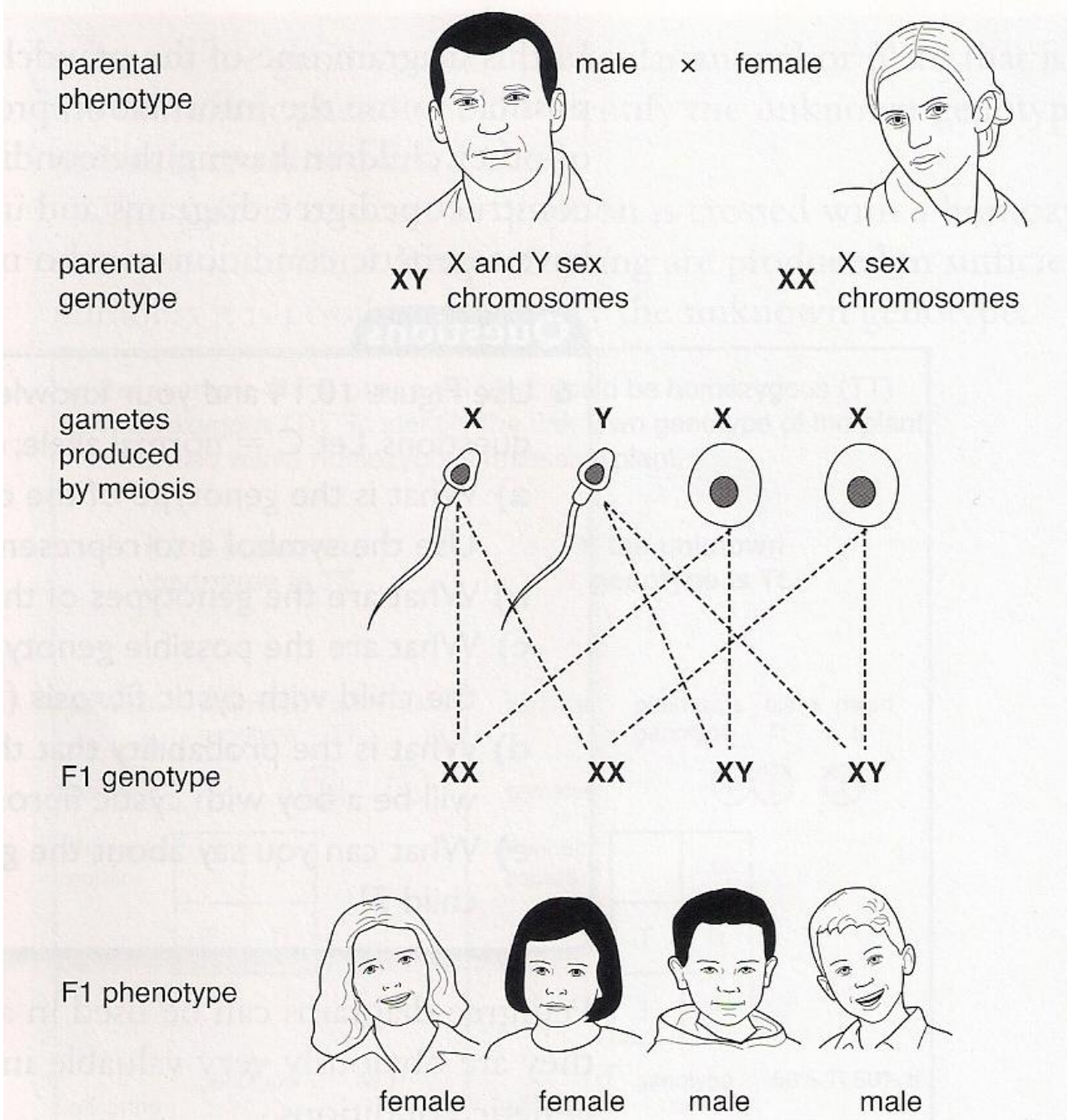
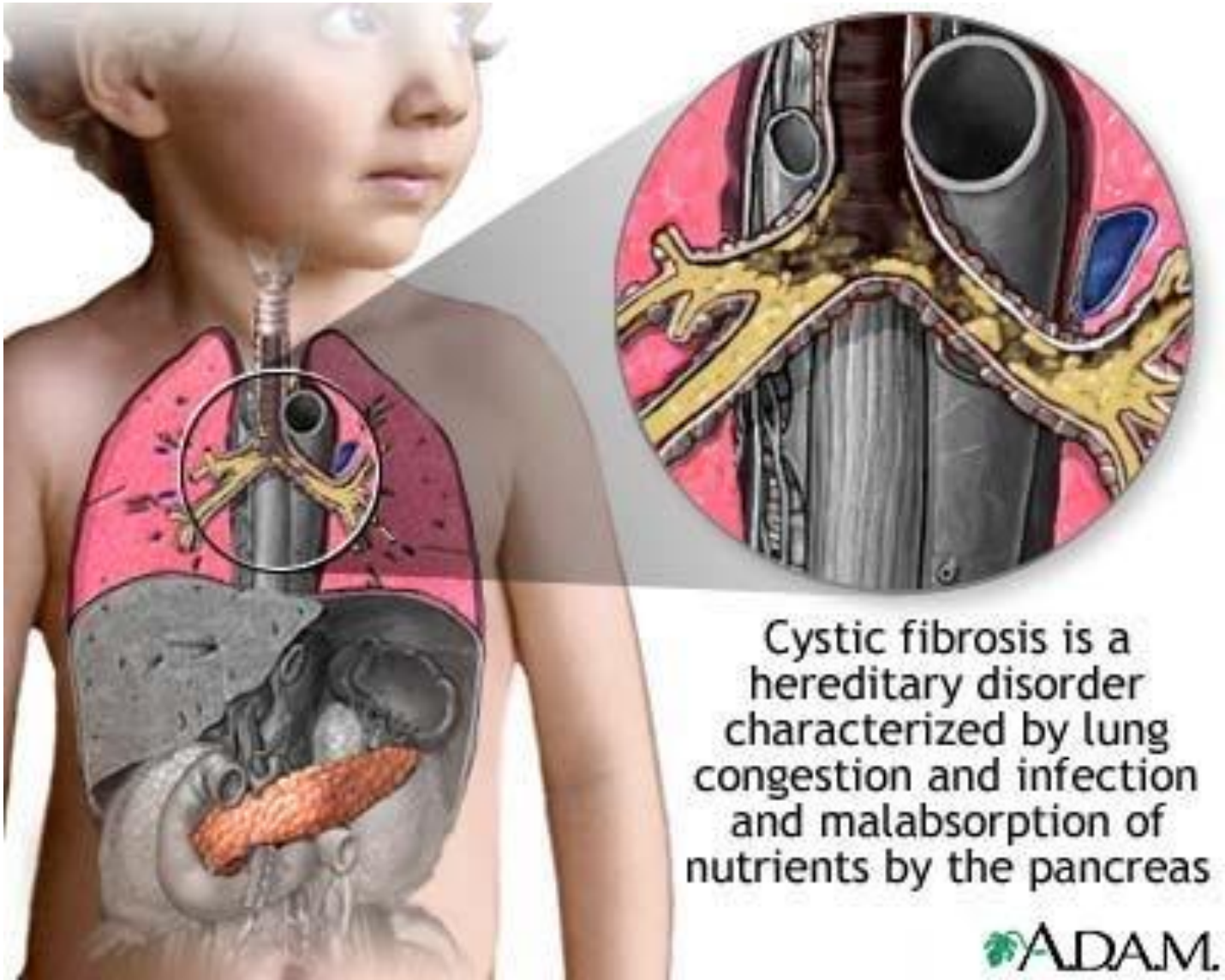


Figure 10.16 Human sex chromosomes

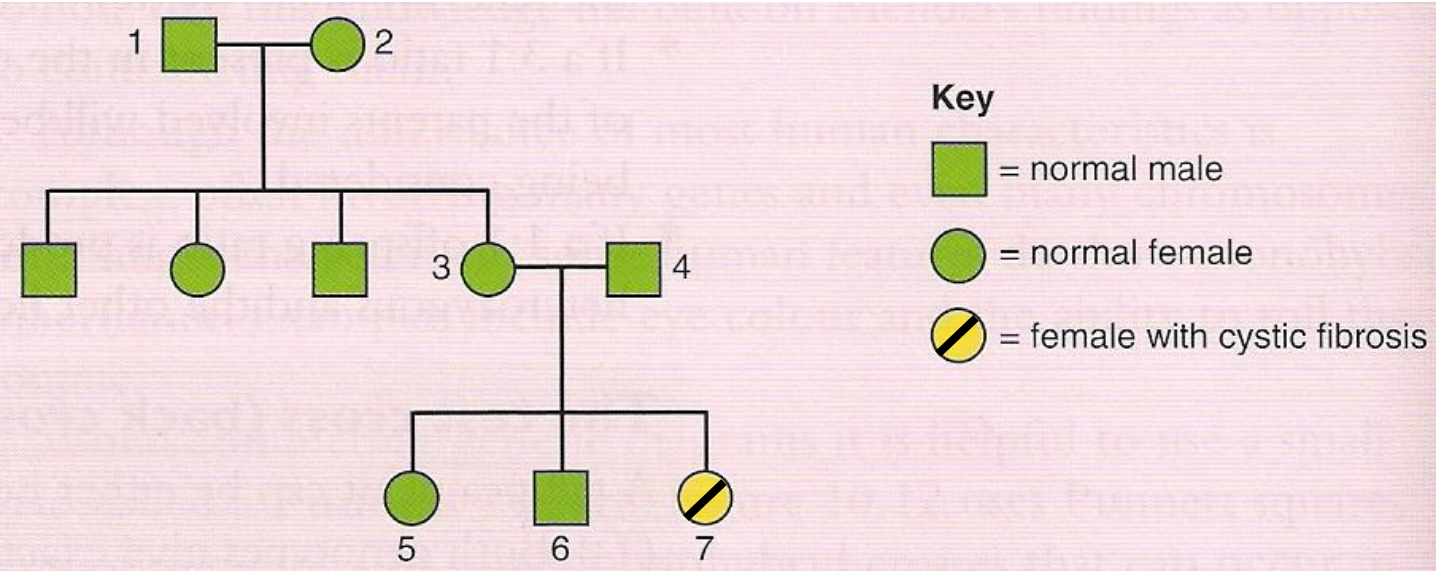
Inherited/Genetic Diseases

Some diseases can be inherited, eg cystic fibrosis which is a medical condition caused by a recessive gene:



Pedigree diagrams:

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? 1

Using the information we can work out the **probability** of other children having the disease. A **genetics counsellor** 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

Questions

6 Use Figure 10.14 and your knowledge to answer the following questions. Let C = normal allele; c = cystic fibrosis allele.

a) What is the genotype of the child (7) with cystic fibrosis? Use the symbol c to represent the cystic fibrosis allele.

b) What are the genotypes of the parents of child 7 (3 and 4)?

c) What are the possible genotypes for the brother and sister of the child with cystic fibrosis (5 and 6)? Explain your answer.

d) What is the probability that the next child of these parents will be a boy with cystic fibrosis?

e) What can you say about the genotypes of the grandparents of child 7?

Answers:

6 a) cc
b) Cc and Cc
c) Cc or CC – they will receive one allele from each parent and they are the possible combinations
d) 0.5 that it will be a boy X 0.25 that it will be homozygous recessive = 0.125
e) It is most likely that one or both will have been heterozygous in order to provide the cystic fibrosis allele to their daughter, the mother of the grandchildren. However, there is a small chance that the cystic fibrosis allele has arisen from a mutation and that the grandparents were both homozygous dominant (CC)

Sex linkage - some diseases called are sex-linked because they are **carried on sex chromosomes**, e.g. inheritance of haemophilia and red-green colour blindness

*Read the following text **slowly and carefully** and answer the questions:*

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?

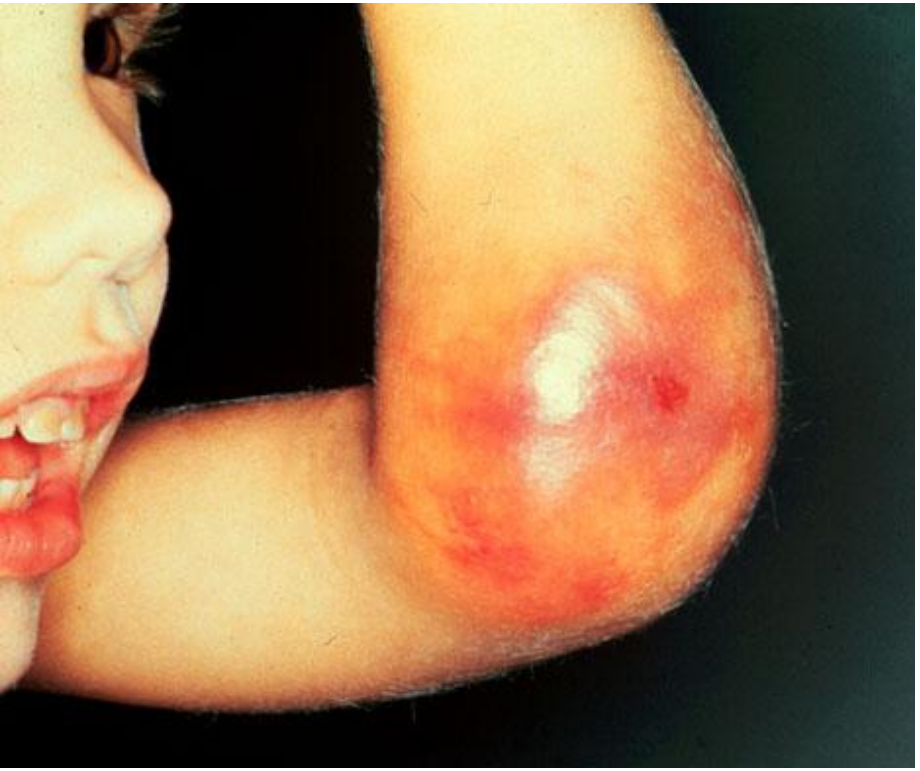
- Why will a recessive allele carried on the X chromosome in a male show its affect in the phenotype?

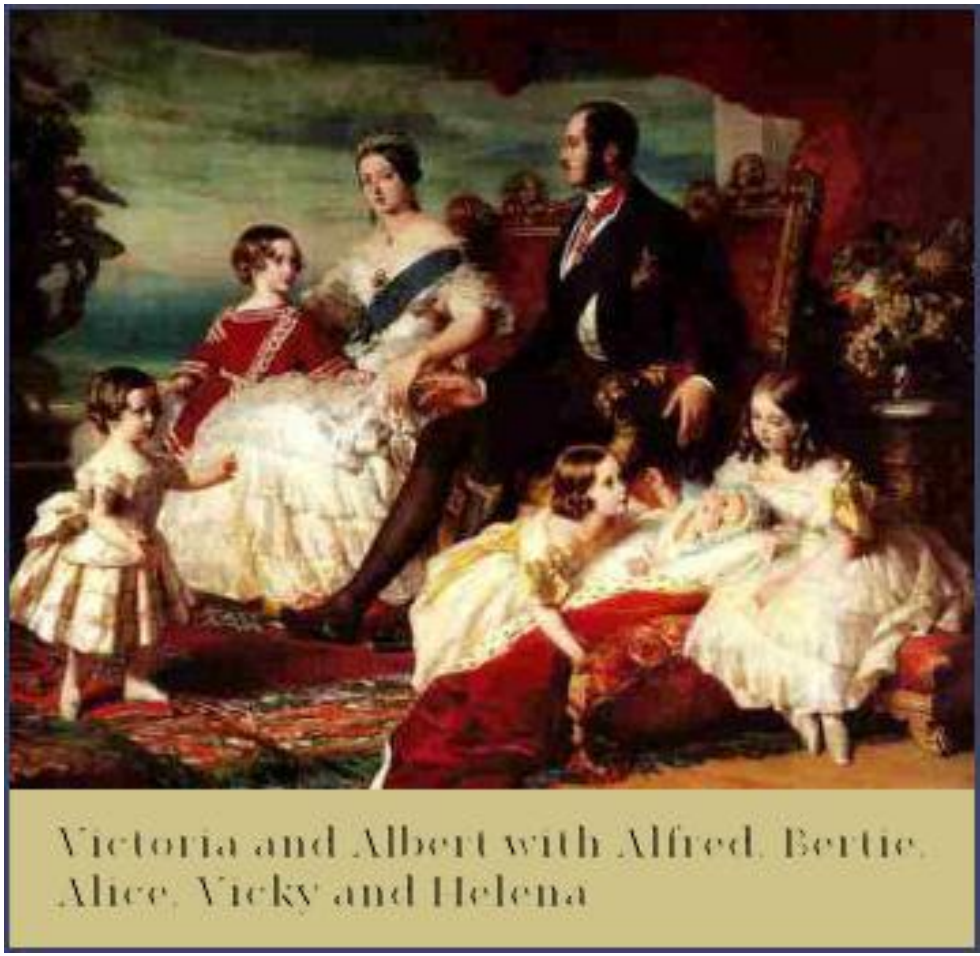
- Although females rarely show sex linked conditions like red-green colour blindness and haemophilia, what role do they often play?

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 **clot** the blood.
Individuals with red-green colour blindness are **unable to distinguish between the colours red and green**

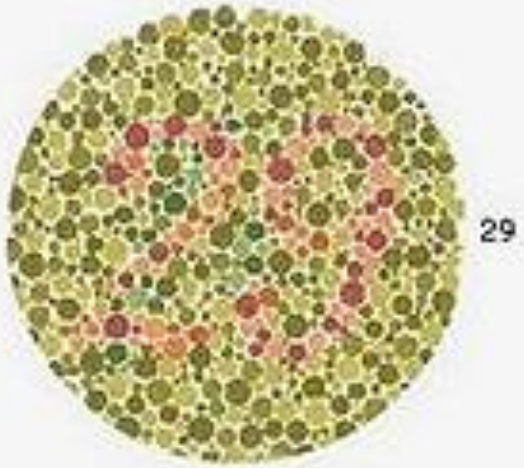
Haemophilia

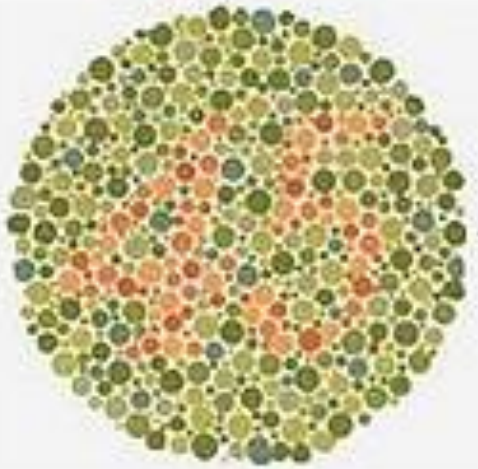


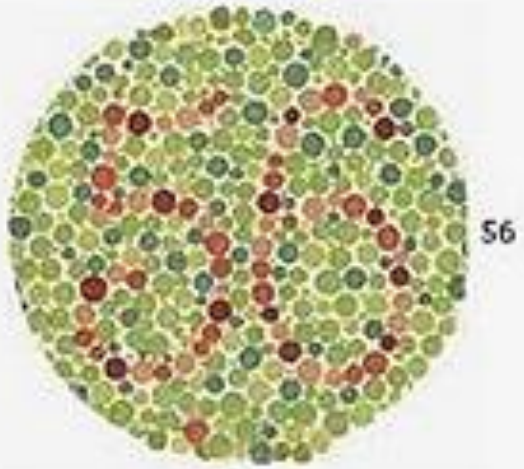


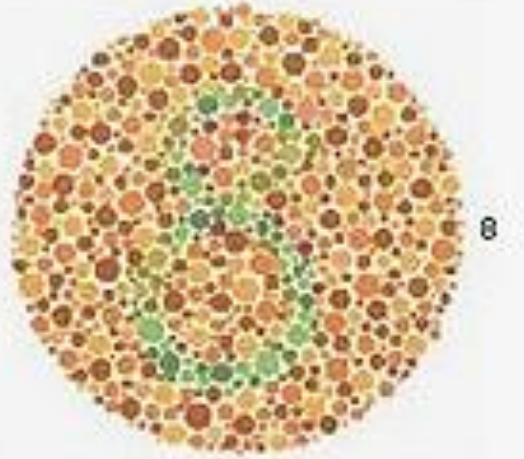
Victoria and Albert with Alfred, Bertie, Alice, Vicky and Helena

Colour Blindness









The inheritance of haemophilia: Complete your diagram

H = normal allele h = haemophiliac allele

Cross 1 haemophiliac male × normal female

parental genotype X^hY X^HX^H

gametes X^h Y X^H

Punnett square

	X^H
X^h	X^HX^h
Y	X^HY

offspring

genotype	50% X^HX^h	50% X^HY
phenotype	50% carrier females	50% normal males

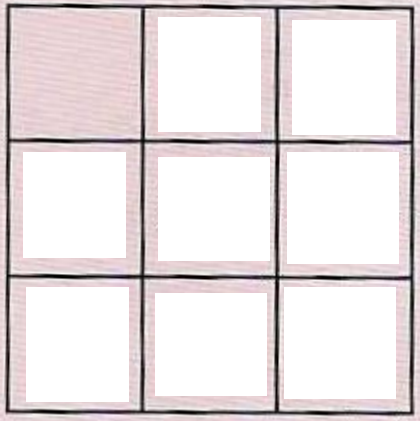
Try and complete cross 2:

Cross 2 normal male × carrier female

parental genotype

gametes

Punnett square



offspring

- genotype: 25% | 25% | 25% | 25%
- 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

ANSWERS:

1 a) Sex linked means that the genes for that disease are carried on the X chromosome

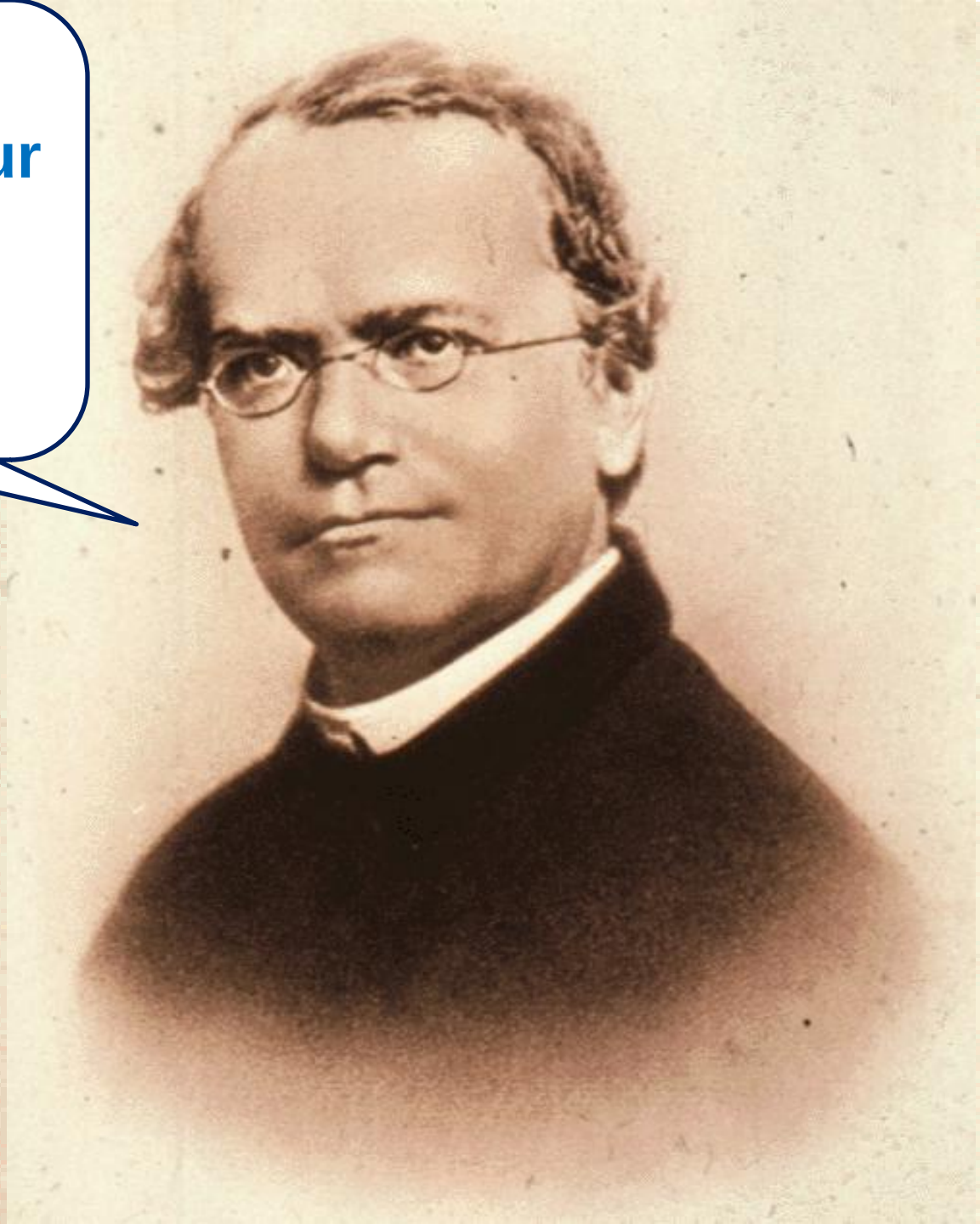
b)

Colour blind man

Carrier woman	X^b	Y
X^B	X^BX^b	X^BY
X^b	X^bX^b	X^bY

25% chance carrier girl
25% chance colour blind girl
25% chance normal boy
25% chance colour blind boy

Revise for your genetics tracking test



HARRY POTTER GENETICS

How are wizards made?

- Being a **wizard** or a **muggle** is all decided by genetics

- All humans including wizards receive one allele from each parent

How are wizards made?

- The allele for **wizarding** ability is **m**

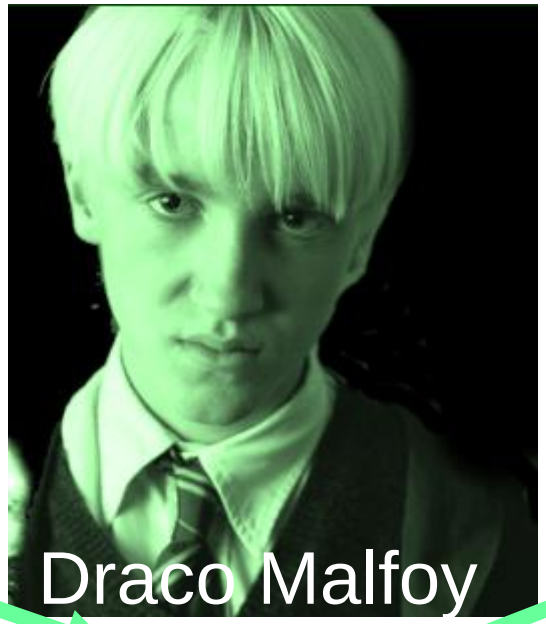
- **Wizards** have the alleles **mm**

How are wizards made?

- The allele for **muggleness** is **M**
- **Muggles** have the alleles **Mm** or **MM**
- **M** is dominant to **m** so you can only be a wizard if you have no **M** allele

THE MALFOYS

Lucius Malfoy
(**mm**)



Draco Malfoy
(**mm**)

Narcissa Malfoy
(**mm**)

The Malfoys are a '**pure blood**' family

All their ancestors are wizards so they must have the alleles **mm**

THE POTTERS

Lily Potter
(**mm**)

Harry Potter
(**mm**)

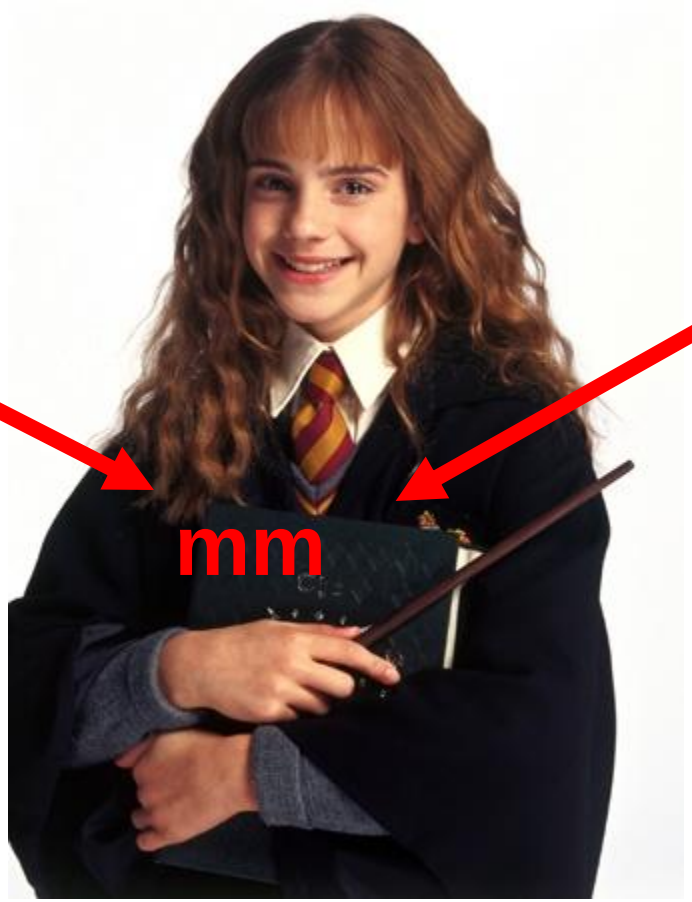
James Potter
(**mm**)

Both Harry's parents had magical ability
so they must both have been **mm**

They passed these alleles on to Harry

The Weasleys are pure blood wizards
so they all have the alleles **mm**

Hermione is a powerful witch so she must be **mm**



Both her parents are **muggles** so they must be **Mm** so they can give her a **m** allele each

Tom Riddle is a '**half blood**'.

His mother was a **witch** (**mm**) and his father was a **muggle**

His father must have had the alleles **Mm** so he could give him the other **m** allele

FILCH IS A 'SQUIB'

Both his parents are **mm** so he should be too because he can't get an **M** allele from either parent but he can't do any magic

This means
either
he has a **mutation** so his wizarding powers don't work
or
the man he thinks is his father isn't really and his mother had an affair with a **muggle**!

What wizarding alleles would Ron and Hermione's children have?

Ron
(**mm**)

Hermione
(**mm**)

Their children could only get the **m** allele from both parents so they would all be wizards

Hermione \ Ron	**m**	**m**
m	**mm**	**mm**
m	**mm**	**mm**

What wizarding alleles would Ginny and Dudley's children have?

If Dudley is **Mm**

Ginny

mm

Dudley

Mm

Half of their children would be likely to get the **m** allele from both parents so they would be **wizards**

The other half would be likely to get an **M** allele from Dudley and would be **muggles**

Dudley

Ginny	**m**	**M**
m	**m m**	**m M**
m	**m m**	**m M**

What wizarding alleles would Ginny and Dudley's children have?

If Dudley is **MM**

Ginny

mm

Dudley

MM

Their children would get the **m** allele from Ginny and the **M** allele from Dudley so they would all be **muggles**

	Dudley	
	M	**M**
Ginny **m**	**m M**	**m M**
m	**m M**	**m M**

What wizarding alleles would you expect Hermione's brothers and sisters to have?

The Granger's children have a one in four chance of getting **m** alleles from both parents and having **magical** ability

They also have a one in four chance of getting **M** alleles from both parents and being a **muggle**

They could also get only one **m** from their mother or father and still be a **muggle**

Mrs Granger

Mr Granger

	m	**M**
m	**m m**	**m M**
M	**m M**	**M M**