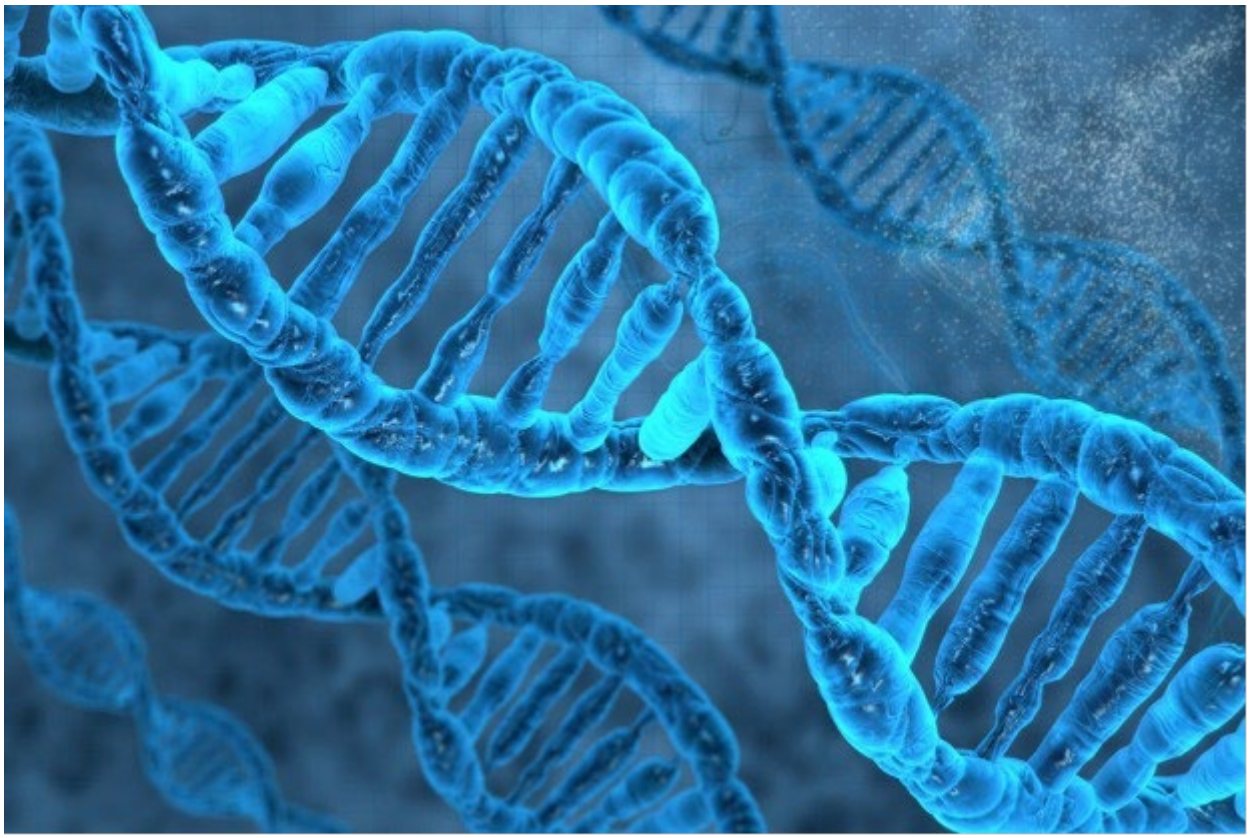
Science Year 10



Medical Emergency
Genetics

This task is continuous of the booklet we're using in the class last term.

Task 1 Watch this YouTube video and read the information on mutation and answer the questions

https://youtu.be/vl6Vlf2thvI

Lesson 6

Mutations

Very occasionally DNA does not replicate perfectly. If there is a **mistake in DNA replication** and the faulty cell survives, it can cause a wide range of problems. The faults that take place during DNA replication are known as **mutations.**
Mutations that occur in **body cells or somatic cells** (example skin cell, bone cells) may affect the organism but may not be inherited and mutation occurring in **gametes (sex cells -eggs (female ovary) sperm (male testes)** will be inherited.
Mutations to DNA can be caused by mutagens such as chemicals, pesticides, tobacco smoke, and by electromagnetic radiation (such as gamma rays, X-rays and UV light).

Diseases caused by mutations include haemophilia, Sickle cell Anaemia, Motor Neuron Disease, Cystic Fibrosis and some cancers.

Types of Mutations

DELETION mutation - a base is lost/deleted

INSERTION -an extra base is added/inserted)

SUBSTITUTION-one base is substituted for another
--- If a substitution **changes** the amino acid, it's called a **MISSENSE** mutation
--- If a substitution **does not change** the amino acid, it's called a **SILENT** mutation
--- If a substitution **changes the amino acid to a "stop**," it's called a **NONSENSE** mutation

Deletion & insertion may cause what's called a FRAMESHIFT mutation, meaning the reading "frame" changes, thus changing the amino acid sequence from this point forward.

Activity: Extension

Complete the boxes below. Classify each as **Deletion**, **Insertion** or **Substitution** AND as either **frameshift**, **missense**, **silent** or **nonsense** (Hint: Deletion & Insertion will always be frameshift).

Original DNA Sequence: T A C A C C T T G G C G A C G A C T …

mRNA Sequence: _____

Amino Acid Sequence:

Mutated DNA **Sequence #1** T A C A T C T T G G C G A C G A C T …

What's the **mRNA** sequence?_____(*Circle the change*)

What will be the **amino acid** sequence? _____

Will there likely be effects? _____ What type of mutation is this? _____

Mutated DNA **Sequence #2** T A C G A C C T T G G C G A C G A C T …

What's the **mRNA** sequence?_____(*Circle the change*)

What will be the **amino acid** sequence? _____

Will there likely be effects? _____ What type of mutation is this? _____

Mutated DNA **Sequence #3** T A C A C C T T A G C G A C G A C T …

What's the **mRNA** sequence?_____(*Circle the change*)

What will be the **amino acid** sequence? _____

Will there likely be effects? _____ What type of mutation is this? _____

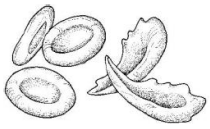
Mutated DNA **Sequence #4** T A C A C C T T G G C G A C T A C T …

What's the **mRNA** sequence?_____(*Circle the change*)

What will be the **amino acid sequence**? _____

Will there likely be effects? _____ What type of mutation is this? _____

Sickle Cell Anemia



Sickle cell anemia is the result of a type of mutation in the gene that codes for part of the **hemoglobin** molecule. Recall that hemoglobin carries **oxygen** in your **red bloods cells**. The mutation causes these red blood cells to become stiff & sickle-shaped when they release their oxygen. The sickled cells tend to get stuck in blood vessels, causing pain and increased risk of stroke, blindness, damage to the heart & lungs, and other conditions.

--- Analyze the DNA strands below to determine what amino acid is changed **AND** what type of mutation occurred

Normal hemoglobin **DNA** C A C G T A G A C T G A G G A C T C …

Normal hemoglobin **mRNA**

Normal hemoglobin **AA** sequence

Sickle cell hemoglobin **DNA** **C A C G T A G A C T G A G G A C A C** …

Sickle cell hemoglobin **mRNA**

Sickle cell hemoglobin **AA** sequence

4. What type of mutation is this? Please explain why.

Activity 2

1. Clarify what is meant by a mutation.

2. What are mutagens?

3. List three mutagens

Lesson 7

Activity 1

Mendel and Inheritance

1.3.1 Gregor Mendel

Gregor Mendel (back row, second from right) is the 'Father of Genetics'.

Gregor Mendel (1822-1884) was an Augustinian monk who investigated inheritance in garden pea plants. He carried out carefully planned experiments breeding peas in the abbey garden. Peas were an excellent choice as an experimental species as he could strictly control which plants mated with which by removing the stamens from plants and then dusting pollen from specific plants onto the stigmas of other specific plants.

Mendel investigated seven characteristics and began with varieties that were **true-breeding**. This means when they self-pollinate all their offspring are that same variety.

Mendel then crossed two contrasting, true-breeding pea varieties, e.g. tall stemmed plants with dwarf stemmed plants and recorded the appearance of all offspring. He called this the **F_1 generation**. He then cross-pollinated these offspring and recorded the appearance of the next generation which he called the **F_2 generation**.

Mendel's results

Characteristic	Traits	F_2 generation	Ratio
Flower colour	Purple / White	705 purple 224 white	3 : 1
Flower position	Axial / Terminal	651 axial 207 terminal	3 : 1
Seed shape	Round / Wrinkled	5474 round 1850 wrinkled	3 : 1
Seed colour	Yellow / Green	6022 yellow 2001 green	3 : 1
Pod shape	Smooth / Constricted	882 smooth 299 constricted	3 : 1
Pod colour	Green / Yellow	428 green 152 yellow	3 : 1
Stem length	Tall / Dwarf	787 tall 277 dwarf	3 : 1

1.3.1.1 What species did Gregor Mendel choose to work with?

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1.3.1.2 Why was this species an excellent choice to study for looking at inheritance patterns?

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.....

1.3.1.3 How did Mendel determine if a plant was true-breeding?

.....

.....

1.3.1.4 How many different characteristics did Mendel study? .....

1.3.1.5 Name one set of contrasting traits for a characteristic. .....

1.3.1.6 When Mendel crossed round seed peas with wrinkled seed varieties, what were his results for the F_2 generation?

.....

7 When Mendel crossed two contrasting, true-breeding pea varieties, he found that the F_1 generation all showed the same trait – which is called the **dominant** trait. He called the other trait the **recessive** trait. When he crossed two of the F_1 generation he found that the dominant trait appeared in a ratio 3 dominant : 1 recessive.

Use the information in the table of Mendel's results to work out which traits in pea plants are dominant and which are recessive.

Dominant traits in pea plants	Recessive traits in pea plants

8 Mendel studied his results and put forward a model of inheritance. He proposed that there were two different traits for each characteristic. One of these traits is dominant while the other trait is recessive. If both traits are present then the dominant trait will show.

Mendel's work was largely ignored at the time and many people believed that inheritance involved a blending of characteristics from one generation to the next.

Choose one of the characteristics of pea plants and propose the results that were needed to show blending.

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9 Explain why Mendel's experiments showed good experimental method.

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10 Mendel published his work in 1866 but it was not until 1900 that his work was rediscovered. Suggest why Mendel is known as the 'Father of Genetics'.

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Activity 2 *Match the vocabulary word with the proper definition.*

Vocabulary words

Allele dominant allele genetics genotype heterozygous

homozygous hybrid law of independent assortment phenotype

law of segregation pollination recessive allele

Definitions

_____ 1. the science of heredity

_____ 2. an organism with two alleles of the same type

_____ 3. an organism with two different alleles

_____ 4. different version of a gene

_____ 5. states that there are two factors controlling a given characteristic, one of which dominates the other, and these factors separate and go to different gametes

_____ 6. states that factors controlling different characteristics are inherited independently of each other

_____ 7. expressed allele in a heterozygote

_____ 8. allele that is not expressed in a heterozygote

_____ 9. the offspring of cross-pollination

_____ 10. fertilization process in the sexual reproduction of plants

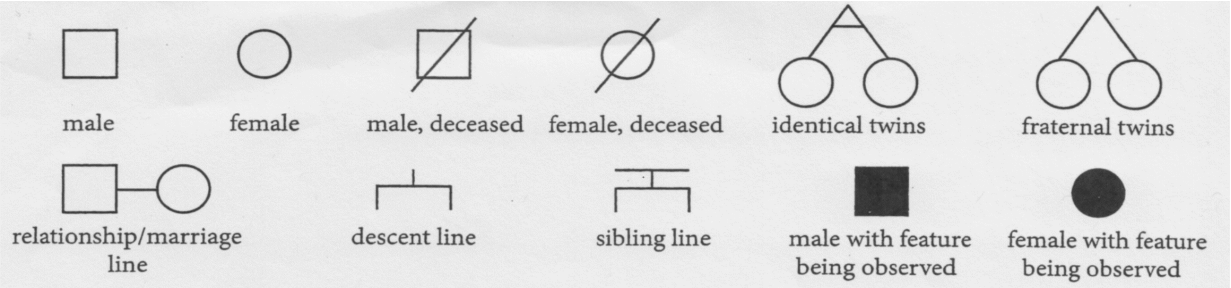
_____ 11. the alleles an individual inherits

_____ 12. the expression of an organism's genotype

Lesson 8 **Pedigree (Family tree)**

Family trees or pedigrees show the inheritance of a particular trait over several generations. Usually only one characteristic is recorded in a pedigree. This characteristic could be the dominant trait or the recessive trait and is indicated by being coloured in. Males are squares and females are circles. The generation is given a roman numeral and each person in the generation has a number.

Symbols used for pedigree



Task 1

Look at the pedigree at the right.

a How many females are there?____

b How many males are there?____

c How many grandchildren are there?____

d Are there any twins in the family?____

e What is the relationship between individuals 7 and 8?_____

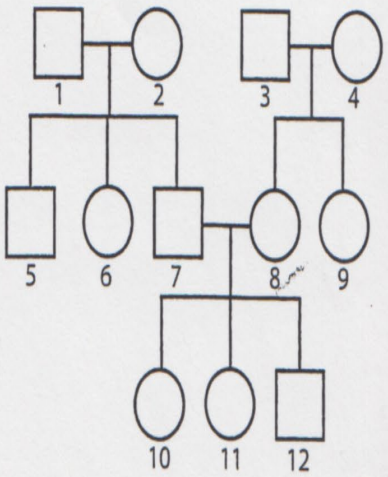
f What is the relationship between individuals 5 and 10?

_____

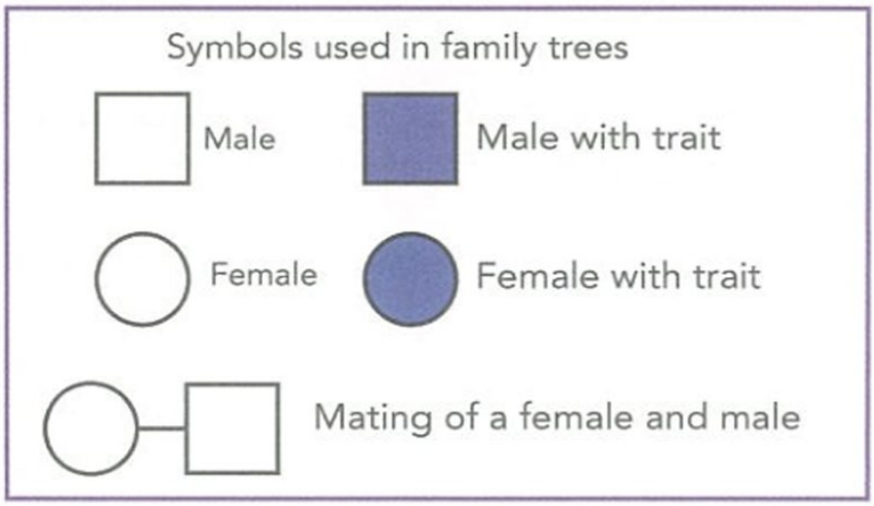
g What is the relationship of individuals 3 and 4 to individual 11?

_____

h All females have the feature of brown eyes. Show that in the pedigree.

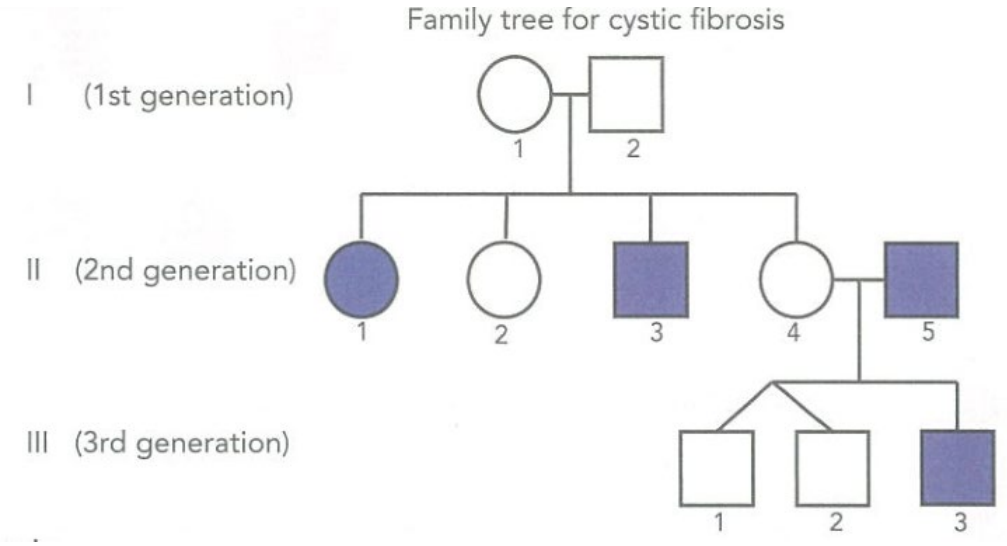


Task 2



Cystic fibrosis is a genetic disease in humans which causes large amounts of mucus to be produced in the lungs. It causes difficulty in breathing and other medical problems.

Cystic fibrosis is caused by a recessive autosomal allele (chromosome linked to body cells or somatic cells not sex chromosomes)



Study the family tree for cystic fibrosis.

1. How many people in this family have cystic fibrosis? _____

2. How do you know they have cystic fibrosis? _____

---

3. How many children did the 1st generation couple (person 1 and person 2) have?

4. Since cystic fibrosis is recessive, a person with this disease would have the genotype ff, while a person without the disease could be FF (homozygous) or Ff (heterozygous) Work out the **genotype** for all members of the family in the pedigree above. (Note that sometimes you cannot tell if a person is FF or Ff if they do not have children of their own.

   I (1st generation): Person 1 _____Person 2 _____

   II (2nd generation): Person 1 _____ Person 2 _____

   Person 3_____ Person 4 _____

   Person 5 _____

   III (3rd generation) Person 1 _____ Person 2 _____

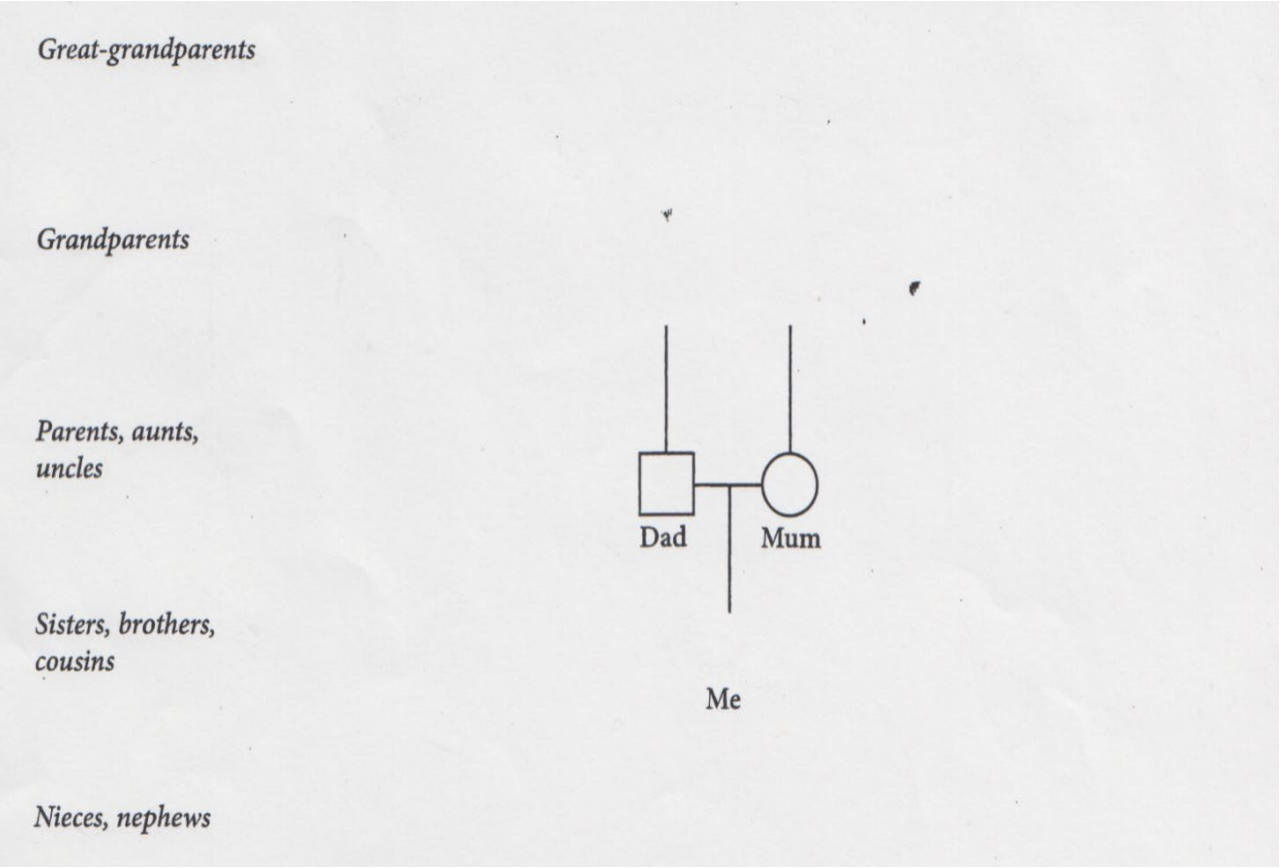
   Person 3 _____

5. Use the Punnett square to work out the probability of couple II4 and II5 having another child with cystic fibrosis.

Task 3

Use the starter pedigree below to draw your own family tree. Use the symbol key provided in the beginning of the lesson.

Choose a trait that runs in the family, like tongue rolling, left handedness, hitch hiker's thumb**.**

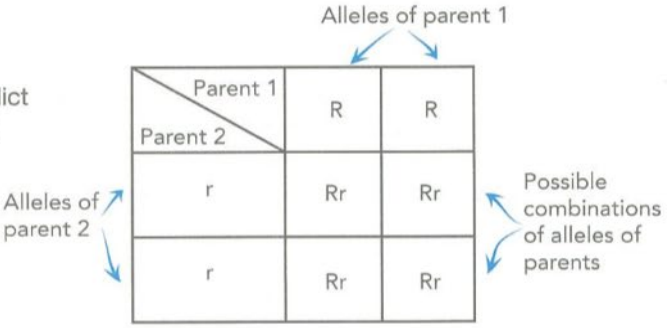


Lesson 9 **Punnet square**

Task 1

1.3.3 Punnett squares

The **Punnett square** is a diagram that is used to predict the outcome of a particular genetic cross. It is named after Reginald Punnett. Punnett squares are used to solve genetics problems. In the square the alleles of the parents are separated to show the possible gametes. When the grid is filled in the possible combinations of alleles in zygotes is shown.



Homozygous and heterozygous

If an organism has identical alleles for a characteristic then it is **homozygous** for that characteristic, e.g. TT for tall stem length in peas or tt for dwarf stem length in peas. If an organism has two different alleles for a characteristic then it is **heterozygous** for that characteristic, e.g. Tt for stem length in peas – this plant would appear tall as the appearance of the dominant allele shows in the phenotype.

1.3.3.1 Who devised the Punnett square as a way to work out the probability of a genetic cross?

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1.3.3.2 Why are Punnett squares used? .....

1.3.3.3 Distinguish between homozygous and heterozygous. .....

.....

1.3.3.4 If a pea plant was heterozygous for each of the following characteristics, what is its genotype and phenotype for each characteristic?

Stem length – heterozygous genotype ..... heterozygous phenotype .....

Flower colour – heterozygous genotype ..... heterozygous phenotype .....

Seed shape – heterozygous genotype ..... heterozygous phenotype .....

1.3.3.5 Draw a Punnett square to show why Mendel found that a cross between a 'true-breeding' (homozygous) tall pea plant and a 'true-breeding' (homozygous) dwarf pea plant always produced tall pea plant offspring.

(*Note:* When solving genetics problems you should always begin with a key to show the code for the dominant and recessive alleles and then show the cross for the parent genotype and phenotypes. Then draw the Punnett square and then write out the genotypes and phenotypes of the offspring in a conclusion.)

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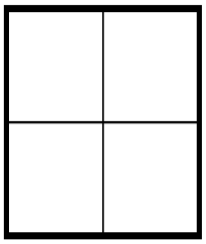
Task 2 More exercises on Punnet squares

Question 1

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!



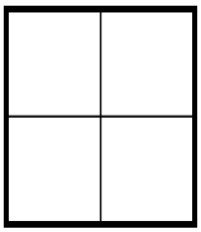
A. List the possible genotypes and phenotypes for their children.

B. What are the chances of a child with a square shape? ____ out of ____ or ____%

C. What are the chances of a child with a round shape? ____ out of ____ or ____%

Question 2

5. Patrick met Patti at the dance. Both of them are heterozygous for their pink body color, which is dominant over a yellow body color. 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? ____ out of ____ or ____%

C. What are the chances of a child with a yellow body? ____ out of ____ or ____%

Question 3

6. Everyone in Squidward's family has light blue skin, which is the dominant trait for body color 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.

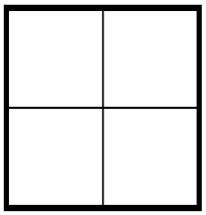
B. What are the chances of a child with light blue skin? ____%

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

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

Question 4

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



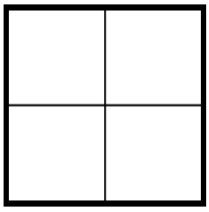
A. List the possible genotypes and phenotypes for their children.

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

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

Question 5

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. Some members of her family have short eyes, which is the recessive trait. 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.

Question 6

4. One of SpongeBob's cousins, SpongeBillyBob, recently met a cute squarepants gal, SpongeGerdy, at a local dance and fell in love. Use your knowledge of genetics to answer the questions below.

(a) If SpongeGerdy's father is a heterozygous squarepants and her mother is a roundpants, what is her genotype? Complete the first Punnett square to show the possible genotypes.

Based on your results, what would Gerdy's genotype have to be? _____

(b) Complete the second Punnett square to show the possibilities that would result if Billy Bob & Gerdy had children.
NOTE: SpongeBillyBob is heterozygous for his squarepantsshape.

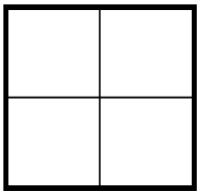
(c) What is the probability of kids with squarepants? _____ %

(d) What is the probability of kids with roundpants? _____ %

Question 7

5. SpongeBob's aunt and uncle, SpongeWilma and SpongeWilbur, have the biggest round eyes in the family. Wilma is believed to be heterozygous for her round eye shape, while Wilbur's family brags that they are a pure line. Complete the Punnett square to show the possibilities that would result if SpongeWilma and SpongeWilbur had children.

(a) Give the genotype for each person. Wilma - _____ Wilbur - _____



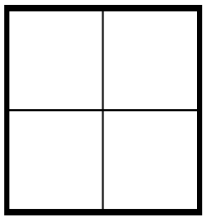
(b) Complete the Punnett square to show the possibilities that would result if they had children.

(c) What is the probability that the kids would have round eyes? ____ %

(d) What is the probability that the kids would be oval eyes? ____ %

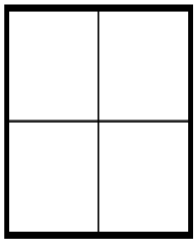
Question 8

6. SpongeBob's mother is so proud of her son and his new wife, SpongeSusie, as they are expecting a little sponge. She knows that they have a 50% chance of having a little roundpants, but is also hoping the new arrival will be blue (a recessive trait) like SpongeSusie and many members of her family. If SpongeBob is heterozygous for his yellow body color, what are the chances that the baby sponge will be blue? Use the Punnett square to help you answer this question.



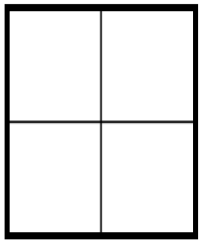
Question 9

7. SpongeBob's aunt is famous around town for her itty, bitty stubby nose! She recently met a cute squarepants fellow who also has a stubby nose, which is a recessive trait. Would it be possible for them to have a child with a regular long nose? Why or why not? Use the Punnett square to help you answer this question.



Question 10

If SpongeBob's aunt described in **question 9** wanted children with long noses, what type of fellow would she need to marry in order to give her the best chances? Use the Punnett square to help you answer this question.



Lesson 10

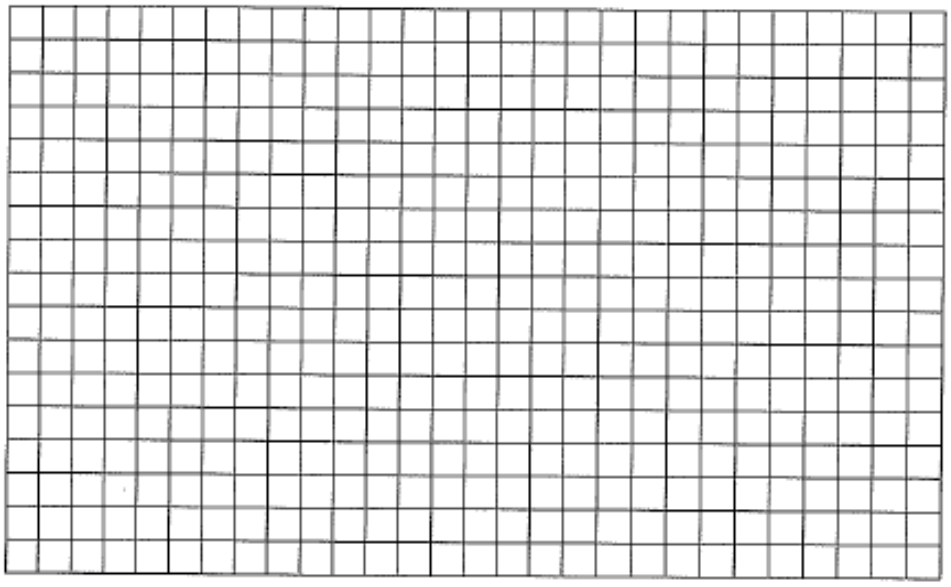
Task 1 Does genetics have to do with me?

Has anyone ever told you that you have your father's nose or your grandmother's hair? Many of our features are inherited from our parents, and theirs from their parents. **Inheritance** or **heredity** is the passing of **characteristics** from parent to offspring – you. Characteristics include eye colour, hair colour, height, nose shape – and many more. We all inherit slightly different combinations of these features from our parents. Even brothers and sisters look different, because they inherit different characteristics from their parents.

1 The table below shows data for characteristics collected from 30 year 9 students. Some of the 30 students did not respond for some characteristics. Add up each tally and calculate the percentage of students with that variation in characteristic. One example has been started for you. *Hint:* The percentages for each variation of a particular characteristic should add up to 100; e.g. 73% + 27% = 100%.

Characteristic	Variations in characteristic	Tally	Tally total	Total number of students	Percentage
Tongue rolling	*Yes, can*	𝍸 𝍸 𝍸 𝍸 ||	22	30	(22 ÷ 30) × 100 = 73%
	No, cannot	𝍸 |||		30	(8 ÷ 30) × 100 = 27%
Attached earlobes	*Yes, attached*	𝍸 𝍸 𝍸			
	No, unattached	𝍸 𝍸			
Receding hair on the sides of the forehead	*Yes, present*	𝍸 𝍸 𝍸			
	No, not present	𝍸 𝍸 𝍸			
Gap between front teeth	*Yes, gap*	𝍸			
	No, no gap	𝍸 𝍸 𝍸 𝍸			
Right thumb on top during hand folding	*Yes, right thumb on top*	𝍸 𝍸 𝍸 𝍸			
	No, left thumb on top	𝍸			
Hair on the second digits of your fingers	*Yes, present*	𝍸 𝍸 𝍸			
	No, not present	𝍸 𝍸			
Right dominant hand	*Yes, right*	𝍸 𝍸 𝍸 𝍸			
	No, left	𝍸			
Second toe longer than big toe	*Yes, longer*	𝍸 𝍸 𝍸			
	No, shorter	𝍸 𝍸 𝍸			
Straight hair	*Yes, straight*	𝍸 𝍸 𝍸 𝍸			
	No, not straight	𝍸 𝍸			

2 Use the percentages you calculated on the previous page to construct a column graph of the results on the grid below.



3 Make two general statements about the graph.

_____

_____

4 Which of the more frequent variations do you possess?

_____

_____

Sometimes characteristics can be influenced by the environment. For example, consider two people with almost identical skin colour – one person likes to be outdoors and the other prefers to spend time indoors. These environments will affect their skin colour. Eating habits can impact on how tall people grow. Poor nutrition can inhibit growth, therefore a person with poor eating habits might not reach full height. Even identical twins can appear different if they are raised in different environments.

5 Which characteristics in the table on the previous page can be altered or influenced by the environment? Explain how.

_____

_____

_____

6 Think about your own development. Do you think it has been influenced more by the environment or more by inheritance? Justify your answer using examples.

_____

_____

_____

Answer page

Task 2 **Is it a boy or a Girl?**

Task 1 Read the information and answer the questions 1 to 8

All organisms are made of cells, and each type of organism has a particular number of chromosomes. For example, each body cell of a dog has 28 chromosomes, while each body cell of a horse has 64 chromosomes. Human cells contain 46 chromosomes, or 23 pairs of chromosomes, or 2 sets of 23 chromosomes (= 2*n*).

Reproductive cells – sperm in males and eggs (ova) in females – always contain half the number of chromosomes of the body cells; that is, one set of 23 chromosomes (= 1*n*). This ensures that the baby also has 46 chromosomes, not 92. When an egg and sperm join during fertilisation, 23 chromosomes come from the mother and 23 come from the father. The fertilised egg has a complete set of 46 chromosomes; that is, 23 pairs of chromosomes.

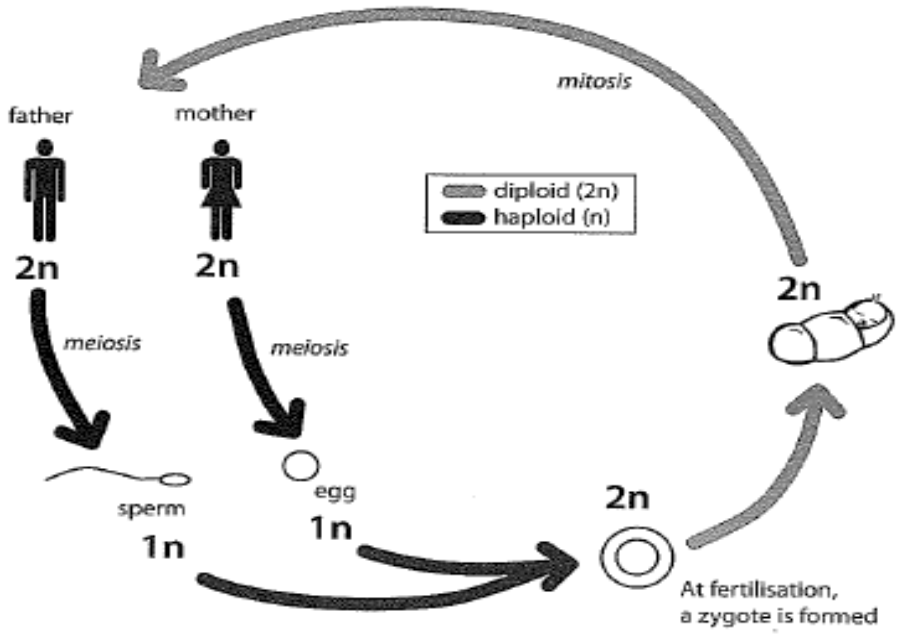


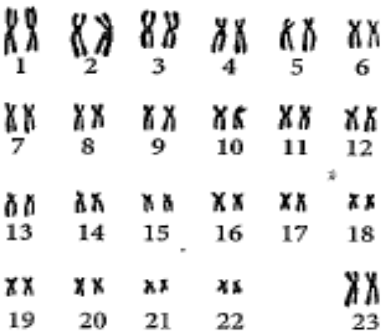
Figure 1

1 What do you think would happen if the egg and sperm always had a complete set of chromosomes?

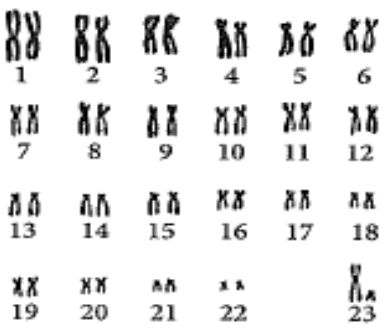
2 Find the terms 'diploid' and 'haploid' in figure 1. What do you think these terms mean?

A special pair of chromosomes determines whether the fertilised egg will be a boy or a girl. This pair is made up of the sex chromosomes. In males, one chromosome in this pair is smaller. The larger chromosome is called X and the smaller is called Y. Females have two X chromosomes, which are the same size. Males have X and Y. These X and Y chromosomes are called sex chromosomes, because they contain the genes that determine sex. The other chromosomes are called autosomes, and these include all the other pairs of chromosomes. Each pair of autosomes in the human karyotype is numbered.

Figure 2 shows the karyotype of a human female and a human male. The first 22 pairs of chromosomes are the autosomes. The 23rd pairs are the sex chromosomes.



Karyotype of a human female



Karyotype of a human male

Figure 2

As shown in figure 3, when a sperm with an X chromosome joins with an egg, the child will be a girl (XX). When a sperm with a Y chromosome joins with an egg, the child will be a boy (XY).

Males produce equal numbers of X and Y sperm, which means the chance of producing a boy or a girl is 50:50.

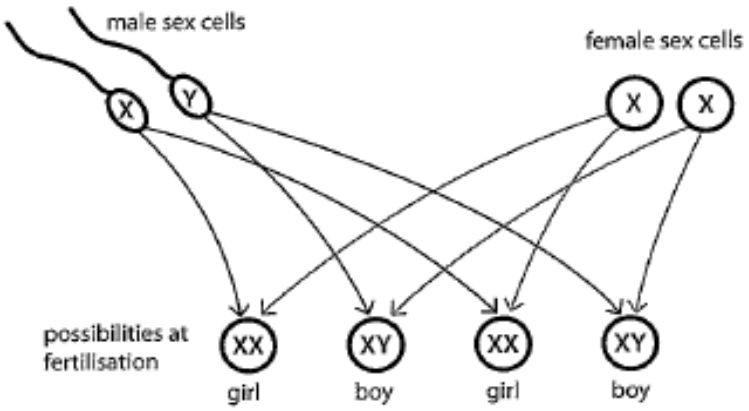


Figure 3

3 Define these terms.

a Autosome **b** Karyotype

4 How many chromosomes do humans have in:

a skin cells? **b** liver cells? **c** sperm?

d ovum? **e** nerve cells?

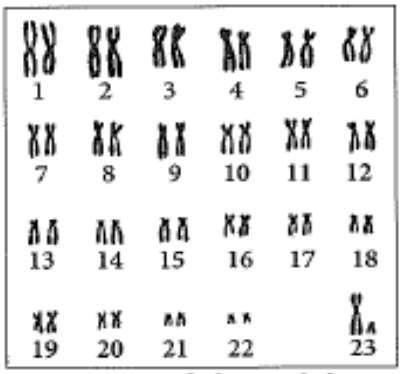
5 Describe how the sex of a human baby is determined.

6 Rewrite the summary points in the table in sentence form.

Location	Cell division	Description	Purpose
Somatic (body) cells	Mitosis	$2n$ (diploid) → $2n$ (diploid)	Growth and repair
Sex organs	Meiosis	$2n$ (diploid) → n (haploid)	Gamete production

7 A karyotype of a human baby is shown at the right.

a What sex is the baby? Explain how you know.
b How many chromosomes does the baby have?
c How many chromosomes did the mother pass on to baby?
d How many chromosomes did the father pass on to baby?
e How many chromosomes are there in the mother's skin cells?
f How many chromosomes are there in the mother's eggs?
g How many chromosomes are there in the father's sperm?
h How many chromosomes are there in the father's skin cells?



Karyotype of a human baby

8 **a** King Henry VIII reigned in England from 1509 to 1547 and had six wives. Historians believe that Henry beheaded his wives when they delivered baby girls, because he wanted a son to continue the royal succession. He blamed his wives for producing daughters instead of sons. Was Henry correct? Who determines the sex of a baby? Explain your answer.

b Why do you think this information was not known at the time of Henry VIII?

Task 3

Define these terms:

Somatic cells –

Diploid-

Mitosis –

Meiosis-

Haploid-

Answer page

Answer page