DNA & INHERITANCE

NAME:

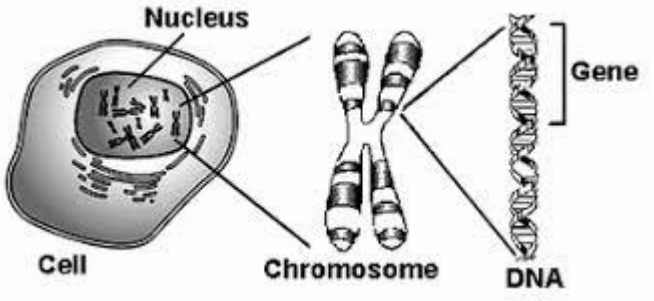
CLASS:

TEACHER:

B6 Inheritance, Variation and Evolution Mastery booklet (COMBINED)

Part 2- DNA and the genome

DNA is often described as the molecule of life. It is responsible for the instructions to make proteins and as such all life's variation and complexity is written in its' DNA. DNA stands for Deoxyribose nucleic acid, and no you don't have to remember that!



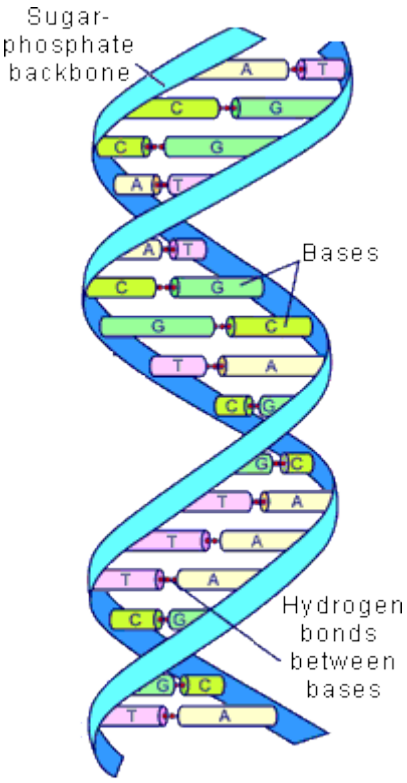
All the DNA is stored as long tightly wound strands called chromosomes. An organisms complete set of genetic material is called its genome.

In 1953 Rosalind Franklin managed to become the first person to prove the structure of DNA. It exists as a **double helix**, formed of two strands which join up and are twisted. DNA is a code. It contains a sequence of **base pairs** which tell the ribosomes the order to place certain **amino acids**. By changing the order of amino acids, different **proteins** are made.

Each **chromosome** is split into sections called **genes**. A gene is a section of DNA which codes for a **protein**. We have now developed the technology to sequence an entire genome. This allows us to know the sequences of bases on each chromosome in an individuals nucleus. The human genome is over 3 billion base pairs long. So far we have used this technology to find genes that link to certain diseases, for example cancer or heart disease. We have also used it to improve our understanding of inherited disorders like cystic fibrosis. The migration of humans throughout history has also been possible. People who share common ancestors also share genes so we can trace the movment of early humans across the globe from their starting point in africe over 1 million years ago. There is hope that this technology will lead to future medical treatments, although as we learn more about the human genome we begin to realise how complex it is and how much more we need to learn.

1. Name the genetic material enclosed in a nucleus of a eukaryotic cell.
2. Where is this genetic material found in a prokaryotic cell?
3. Describe the structure of double helix.

4. Name the structure the genetic material organised itself into.
5. Define ‘gene’.
6. Define ‘genome’.
7. How many base pairs are found in the human genome?
8. Name 3 uses for the sequenced human genome?
9. What organelle makes proteins from amino acids?



Part 3- Genetic inheritance

Individuals within a species all display variation. This is due to **sexual reproduction**. In sexual reproduction **gametes fuse** to form a unique offspring. They are unique because they have inherited half their **chromosomes** from their father and half from their mother. Each time a new offspring is conceived a different combination of the chromosomes combine; this is why you will resemble your brother/sister but are not identical to them.

Different forms of the same gene are known as **alleles**.

There are 2 copies of every chromosome in a body cell nucleus (1 copy inherited from the mother the other copy inherited from the father). Therefore, there are 2 copies of every gene. These copies may be different alleles and the combination of the 2 alleles (the **genotype**) determines the characteristic (the **phenotype**).

If the 2 alleles for a gene are the same, we call this combination **homozygous**, whereas if the 2 alleles are different, we call this combination **heterozygous**.

For most genes, one allele is said to be **dominant** and the other **recessive**. If a dominant allele is present, only its phenotype is expressed. This means individuals that have 2 dominant alleles (**homozygous dominant**) OR one dominant and one recessive allele (**heterozygous**) will express the dominant phenotype. Both alleles need to be recessive (**homozygous recessive**) for the recessive phenotype to be expressed. Due to this relationship, we often refer to the alleles using the same letter, the dominant in UPPER case and the recessive in lower case e.g. “B” is dominant, “b” is recessive.

For example: Consider a flowering plant that can have pink or white petals. Pink is the dominant allele, so our code is P= pink and p=white

- PP is the homozygous dominant, its phenotype is pink.
- Pp is heterozygous, its phenotype is also pink as pink is dominant to white.
- pp is homozygous recessive; its phenotype is white.

In our work we will be looking at characteristics that are controlled by one gene. In reality multiple genes work together to create the overall phenotype. A good analogy is that of an orchestra, with many different instruments working in harmony to create the music.

10. Define all the terms in **bold**
    The gene for flower colour in pea plants has 2 alleles, purple and white. The purple allele is represented by “R” and the white allele by “r”.
11. What is the homozygous dominant genotype?
12. What is the homozygous dominant phenotype?
13. What is the homozygous recessive genotype?
14. What is the homozygous recessive phenotype?
15. What is the heterozygous genotype?
16. What is the heterozygous phenotype?

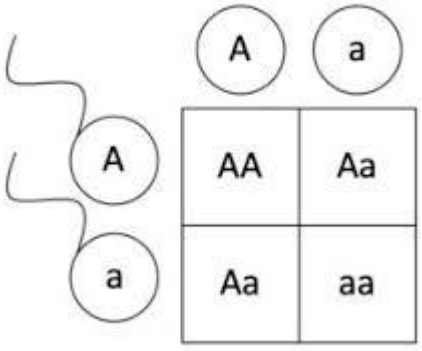
17. Complete the table below. The first one has been done for you.

	Homozygous dominant		Homozygous recessive		Heterozygous	
Scenario	genotype	phenotype	genotype	phenotype	genotype	phenotype
A. The gene for eye colour in humans has 2 alleles, blue and brown. The brown allele is represented by "B" and the blue allele by "b".	BB	Brown	bb	Blue	Bb	Brown
B. The gene for plant height in sunflowers has 2 alleles, tall and dwarf. The dwarf allele is represented by "d" and the tall allele by "T".						
C.The gene for flower position in courgettes has 2 alleles, terminal (F) and axial (f).						
D.The gene for coat colour in cows has 2 alleles, roan (g) and white (G).						

Punnett squares.

When examining the inheritance of alleles through multiple generations we use genetic cross diagrams or punnett squares.

Below is the punnett square for the inheritance of recessive condition called Albinism.



18. Label the punnett square with the following labels: *alleles from father, alleles from mother, possible genotypes*
19. What proportion of the possible genotypes is homozygous dominant?
20. What proportion of the possible genotypes is heterozygous?
21. What proportion of the possible genotypes is homozygous recessive?

22. What proportion of the possible offspring will be healthy?
23. What proportion of the possible offspring will have Albinism?

The gene for flower colour in pea plants has 2 alleles, purple and white. The purple allele is represented by “R” and the white allele by “r”.

For each of the questions below:

a) Draw a Punnett square to show the 4 possible offspring genotypes from this breeding.
b) For each offspring, label the phenotype.
c) For each offspring, describe the genotype using the words homozygous, heterozygous, dominant and recessive.
d) Calculate the probability of each phenotype.

24. Two pea plants both with the genotype Rr breed.
25. Rr x rr
26. RR x rr
27. Rr x RR

Part 3- Inherited disorders

Sometimes a mutation happens in the chromosomes of a gamete. In this case the faulty gene will be present in every cell of the body. In some very rare cases this can cause an inherited disorder. The disorder is inherited because there is a chance it can be passed down to the next generation. Some inherited disorders are dominant and other are recessive, we will look at an example of both.

Polydactyly

Polydactyly is an inherited disorder that results in the child growing extra fingers or toes. It does not have any significant long term health problems. It is caused by a **dominant allele**. This means that only one parent needs to pass on the faulty allele for the child to suffer from polydactyly, So a **homozygous dominant** parent is guaranteed to have a polydactyly child and a **heterozygous** parents will have a chance of having one.

Picture 1
Normal Hand

Picture 2
Hand with Polydactyly

To the right is a punnett square showing the chances of having child with polydactyly when one parent with the disorder mates with a homozygous recessive partner.

P	PP 25%	Pp 25%
p	Pp 25%	pp 25%

In this example:
Genotypes

- 25% chance of the homozygous dominant PP
- 50% chance of the heterozygous Pp
- 25% chance of the homozygous recessive

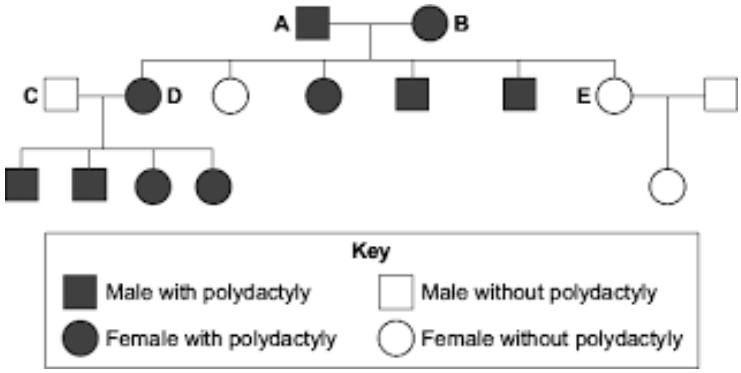
Phenotypes

- 75% chance of having polydactyly (as it is dominant)
- 25% chance of being unaffected

28. Using the example above, draw a punnett square to show the outcomes of the following crosses:
    a) Pp x Pp
    b) A heterozygous father and a homozygous recessive mother

29. Suggest why polydactyly is not considered a serious inherited disorder

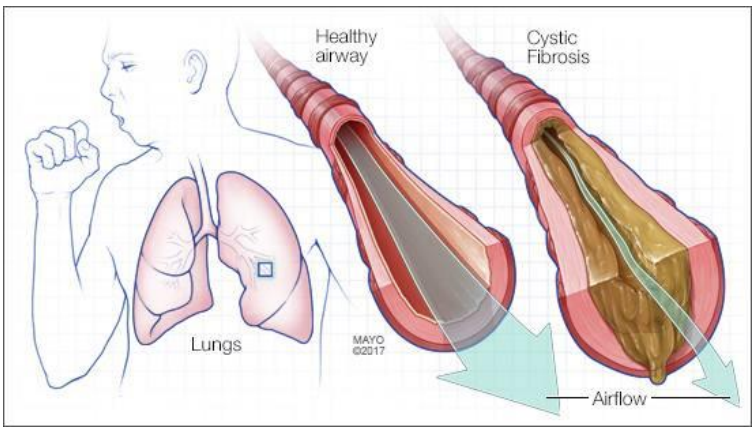
Another way of determining the dominant nature of polydactyly is through a **family tree** diagram. The clue to its dominant nature is the breeding between D and C. As their children all suffer from the disease there is a very strong chance the condition is dominant.



Cystic Fibrosis

Cystic fibrosis is an inherited disorder which affects the mucus that lines the respiratory and digestive tract. Cystic fibrosis sufferers have much thicker mucus than normal. This makes breathing harder and increases their chance of chest infections. Their pancreatic duct can also be blocked, resulting in less digestive enzymes making their way into the small intestine. This can affect the amount of nutrients they get from their food. Currently the disorder is managed using a combination of physical therapy (to remove the mucus) and drugs (to improve the digestion). A more long term solution is for a heart and lung transplant.

Cystic fibrosis is caused by a recessive allele. This means that only **homozygous recessive** people are suffering from the disorder and **heterozygous** people are completely normal, but they have a chance of having a child which suffers from it. They are known as **carriers**. In the uk it is estimated that 1 in 25 people is a heterozygous carrier. Currently there are approximately 10,500 people with cystic fibrosis in the UK. This is about 1 in every 2500 babies born.



To the right is the punnett square for a cross between two heterozygous carriers. C= healthy, c= cystic fibrosis

	C	c
C	CC	Cc
c	Cc	cc

In this example:

Genotypes

- 25% chance of the homozygous dominant CC
- 50% chance of the heterozygous Cc
- 25% chance of the homozygous recessive cc

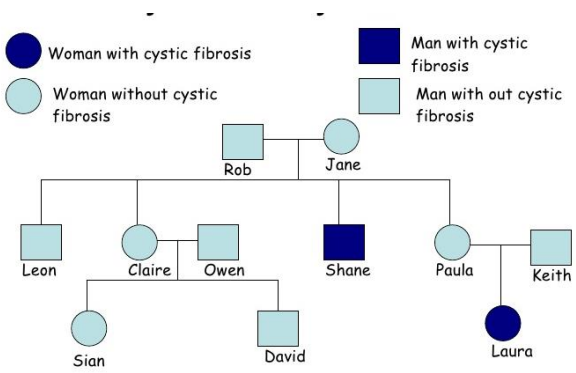
Phenotypes

- 25% of being unaffected CC
- 50% of having no symptoms but being a carrier Cc

- 25% chance of suffering from cystic fibrosis cc

If we look at the family tree a key clue to it being a recessive condition can be seen.

Recessive inherited conditions have the ability to skip generations. Consider Rob and Jane. Both did not suffer from CF, but had a child (Shane) who did. This pattern is also repeated with Paula and Keith. In both cases it must be that they were both heterozygous carriers, a key defining factor of a recessive inherited condition.



30. Using the example above, draw a punnett square to show the outcomes of the following crosses:
    c) CC x cc
    d) A heterozygous father and a homozygous recessive mother
31. If 1 in 25 people are carriers suggest why so few babies are born with cystic fibrosis?
32. If the population of the UK is 6.6 x 10^6 people how many people are carriers? Give your answer in standard form.

Genetic tests

Genetic testing involves analysis of a person's DNA to see if they carry alleles that cause genetic disorders. It can be done at any stage in a person's life.

- **Antenatal testing** is used to analyse an individual's DNA or chromosomes before they are born. This testing is offered to couples who may have an increased risk of producing a baby with an inherited disorder, but it can't detect all the risks of inherited disorders.
- **Neonatal testing** known as the new born blood spot test involves analysing a sample of blood that is taken from pricking a baby's heel. It detects genetic disorders in order to treat them early.
- **Pre-implantation genetic diagnosis (PGD)** is also called **embryo screening.** It is used on embryos before implantation. Fertility drugs stimulate the release of several eggs. The eggs are collected and fertilised in a Petri dish. This is known as in vitro fertilisation (IVF). Once the embryos have reached the eight-cell stage, one cell is removed.

The cells are tested for the disorder causing alleles. Embryos that don't contain the disorder allele are implanted into the uterus.

Limits of genetic testing

Genetic tests are not available for every possible inherited disorder, and are not completely reliable. They may produce false positive or false negative results, which can have serious consequences for the parents involved.

False positives

A false positive is a genetic test that wrongly detected a certain allele or faulty chromosome. The individual could believe something is wrong, when it is actually fine.

False negatives

A false negative is a genetic test has failed to detect a certain allele or faulty chromosome. The parents may be wrongly reassured. These results can have a major impact on the lives of individuals, through pregnancy termination, future decisions and planning the level of care needed for children with inherited disorders.

On top of the technical problems, people can be against these tests for moral and spiritual reasons. Most of these stem from a strong belief that it is wrong to tamper with the natural process of reproduction. With embryo screening techniques some of the embryos are destroyed. Some people believe that this is the same as murder and so are against this process.

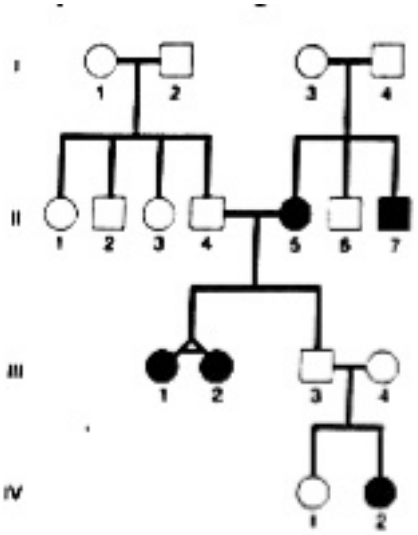
33. What is the meaning of 'inherited disorder'?
34. Mariana says "Measles is an inherited condition because my dad had it when he was young and now I've had it!" Explain why she is wrong.
35. What medical treatment could Mariana's parents have given her when she was young to prevent her catching measles?
36. Explain the difference between a dominant and recessive inherited disorder.
37. What are the symptoms of poydactyly?
38. What are the symptoms of cystic fibrosis?
39. What can you not be a carrier of polydactyly?
40. What three ways are inherited disorders tested for?
41. Why is every baby born given the heel prick test (neonatal), but only high risk pregnancies given antenatal testing?
42. Why would a person who is against abortion refuse PGD?
43. What is the difference between a false positive and a false negative? Which do you think is worse?

Huntington's disease is a dominant inherited condition. It causes problems in a persons' ability to use their muscles, including breathing and swallowing. There is no cure. Symptoms begin to show during a person's 30's and 40's.

44. Can you be a carrier for huntington's?
45. Will the huntington's allele be H or h?
46. Draw a punnett square for a heterozygous and homozygous recessive breeding. Give the proportions of each genotype and phenotype.

Thalassemia is an inherited disorder. It causes the red blood cells to twist into a different shape. This affects the person's ability to get oxygen to their organs, causing fatigue, and a yellowing of the skins.

47. Look at the family tree for Thalassemia. Is it a dominant or recessive inherited condition? Give a reason.
48. Draw a punnett square for a heterozygous and a homozygous recessive. Give the proportions of the genotypes and the phenotypes.



49. In humans, chromosome **X** and chromosome **Y** are the sex chromosomes.

    (a) Most cells in the human body contain two sex chromosomes. Which type of cell does **not** have two sex chromosomes? Choose from Liver cell, Nerve cell, Red blood cell

    (b) Apart from the sex chromosomes, how many **other** chromosomes are there in most human body cells?

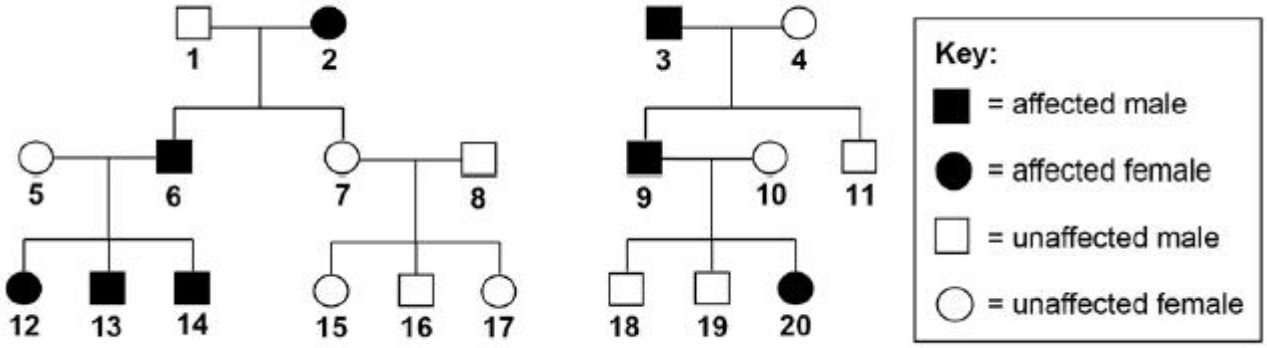
Stickler syndrome is an inherited disorder that causes damage to the eye.

One of the symptoms of Stickler syndrome is that black spaces can appear in the visual image.

(c) Which part of the eye is affected by Stickler syndrome?

Stickler syndrome is caused by the inheritance of a dominant allele.

The diagram shows the inheritance of Stickler syndrome in two families.



Use the following symbols in your answers to (d) and (e):

A = the dominant allele for Stickler syndrome

a = the recessive allele for unaffected vision.

(d) Explain why none of the children of persons **7** and **8** have Stickler syndrome.

(e) Person **12** marries person **18**.

Use a Punnett square diagram to find the probability that their first child will be a female with Stickler syndrome.

Part 3 – Sex determination

Ordinary human body cells contain 23 pairs of chromosomes. 22 pairs control characteristics only, but one of the pairs carries the genes that determine sex.

• In females the sex chromosomes are the same (XX).

• In males the chromosomes are different (XY).

In the punnett square it shows the possible gender outcomes of sexual reproduction.

	X	X
X	XX	XX
Y	XY	XY

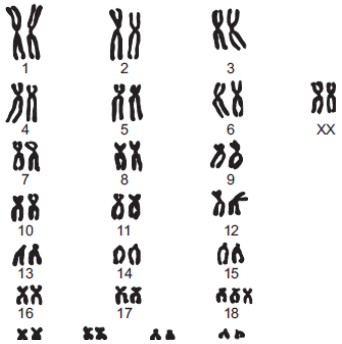
50. What is the percentage chance a baby is female.
51. What re the possible genotypes of the sperm?
52. Genetic disorder **E** is a condition caused by a change in the chromosomes.

(a) **Figure 1** shows the chromosomes from one cell of a person with genetic disorder **E**.

(i) How do you know this person is female? Use information from **Figure 1**.

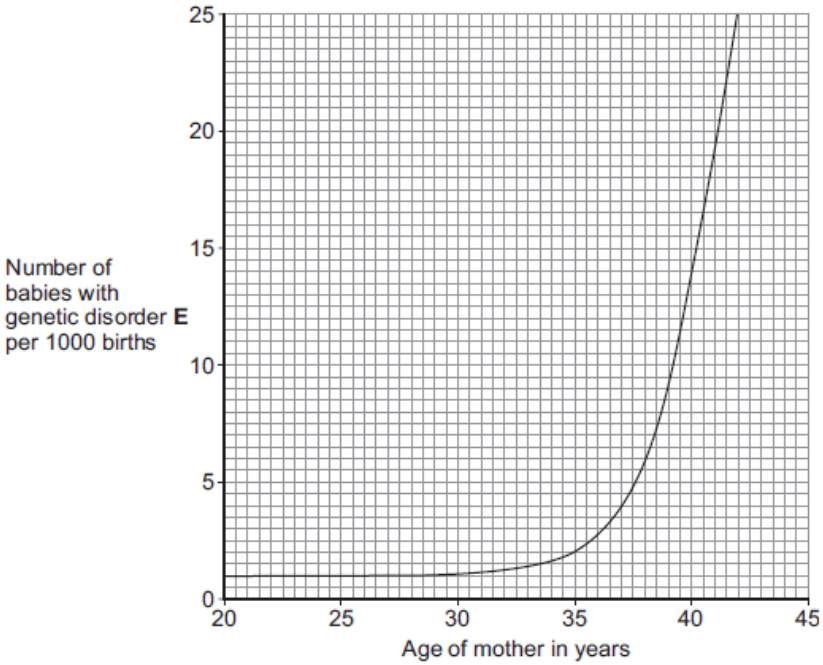
(ii) Describe how the chromosomes shown in **Figure 1** are different from the chromosomes from a person who does not have genetic disorder **E**.

Figure 1



(b) As a woman gets older, the chance of her having a baby with genetic disorder **E** increases. **Figure 2** shows this.

Figure 2



(i) The chance of a 35-year-old woman having a baby with genetic disorder **E** is 2 per 1000 births.

What is the chance of a 40-year-old woman having a baby with genetic disorder **E**?

(ii) A 40-year-old woman is more likely than a 35-year-old woman to have a baby with genetic disorder **E**. How many times more likely?

(c) A 41-year-old woman wants to have a baby. A 41-year-old woman has an increased chance of having a baby with genetic disorder **E**. Doctors can screen embryos for genetic disorder **E**. The table gives some information about two methods of embryo screening.

Method 1	Method 2
1. The woman is given hormones to cause the release of a few eggs. The eggs are taken from her body in a minor operation. The eggs are fertilised in a glass dish.	1. The woman gets pregnant in the normal way.
2. One cell is taken from each embryo when the embryo is 3 days old.	2. Cells are taken when the embryo is 10 weeks old.
3. Cells are screened for genetic disorder **E**.	3. Cells are screened for genetic disorder **E**.

4. An unaffected embryo is placed in the woman's uterus. Embryos that are not used are destroyed or used in medical research.	4. An unaffected fetus is allowed to develop. If the fetus has genetic disorder **E**, the woman can choose to have an abortion.
5. This method costs about £6000.	5. This method costs about £600.

Use information from the table to give **two** advantages and **one** disadvantage of **Method 1**compared with **Method 2** for detecting genetic disorder **E**.