10 F Science
Genetics and Species Survival Part 1: Task Booklet 2

You are a researcher investigating heart disease. You know there are 6 genes in humans that can contribute to heart disease risk. All humans have these 6 genes, but we can inherit slightly different forms or "flavors" of these genes. Your challenge is to track and record the passage of these 6 genes (signified by colored pom poms) through generations of a family using a pedigree. Then, predict which members of this family are most likely to develop heart disease.

Follow the steps below to complete this challenge.

Part 1:

1. Obtain a blank pedigree, colored pencils or crayons, 2 disposable cups, and an assortment of colored pom poms from your instructor.
2. Choose one cup that will represent a grandmother. Place inside the cup 5 red pom poms (5 "normal" genes that do NOT contribute to heart disease) and 1 blue pom pom (1 gene that increases heart disease risk).
3. Choose one cup that will represent a grandfather. Place inside the cup 3 red pom poms (3 "normal" genes that do NOT contribute to heart disease), as well as 1 orange, 1 green, and 1 yellow pom pom (3 genes that increase heart disease risk).
4. On the pedigree, record the colors of pom poms present in both the grandmother and grandfather by filling in the blank circles using crayons or colored pencils.
5. With closed eyes, mix and randomly draw out 3 pom poms from each grandparent (6 pom poms total). This will represent the genetic information inherited by their first child.
6. On the Pedigree, color in the combination of pom poms that were passed on to the child.
7. Return the pom poms to the appropriate grandparent. (Give grandma's genes back to grandma, and give grandpa's genes back to grandpa.)
8. Repeat steps 5-7 for each son or daughter in the second row of the pedigree. Do not do the son's partner.

9. Diagnosis:

Look at each individual's 6 heart disease susceptibility genes (their combination of colored pom poms). Remember: The forms or "flavors" of a gene that increase heart disease risk are

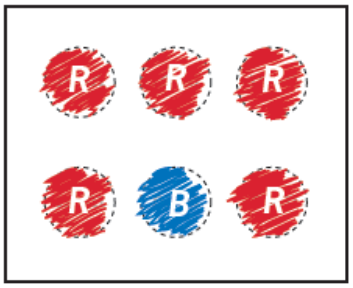
yellow(Y): **orange(O):** **green(G):** **blue(B):**

Label each individual in your pedigree as low, medium or high risk according to the chart below.

High Risk

Medium Risk

Low Risk



Part 2:

10. Place in a cup 6 pom poms that are the colors you drew for the second son in row 2.

11. The partner of the second son is a "low risk" woman. Place inside a cup 5 red pom poms and 1 yellow pom pom to represent her 6 heart disease susceptibility genes.

12. On the pedigree, color in the combination of pom poms (or genes) carried by the partner of the second son.

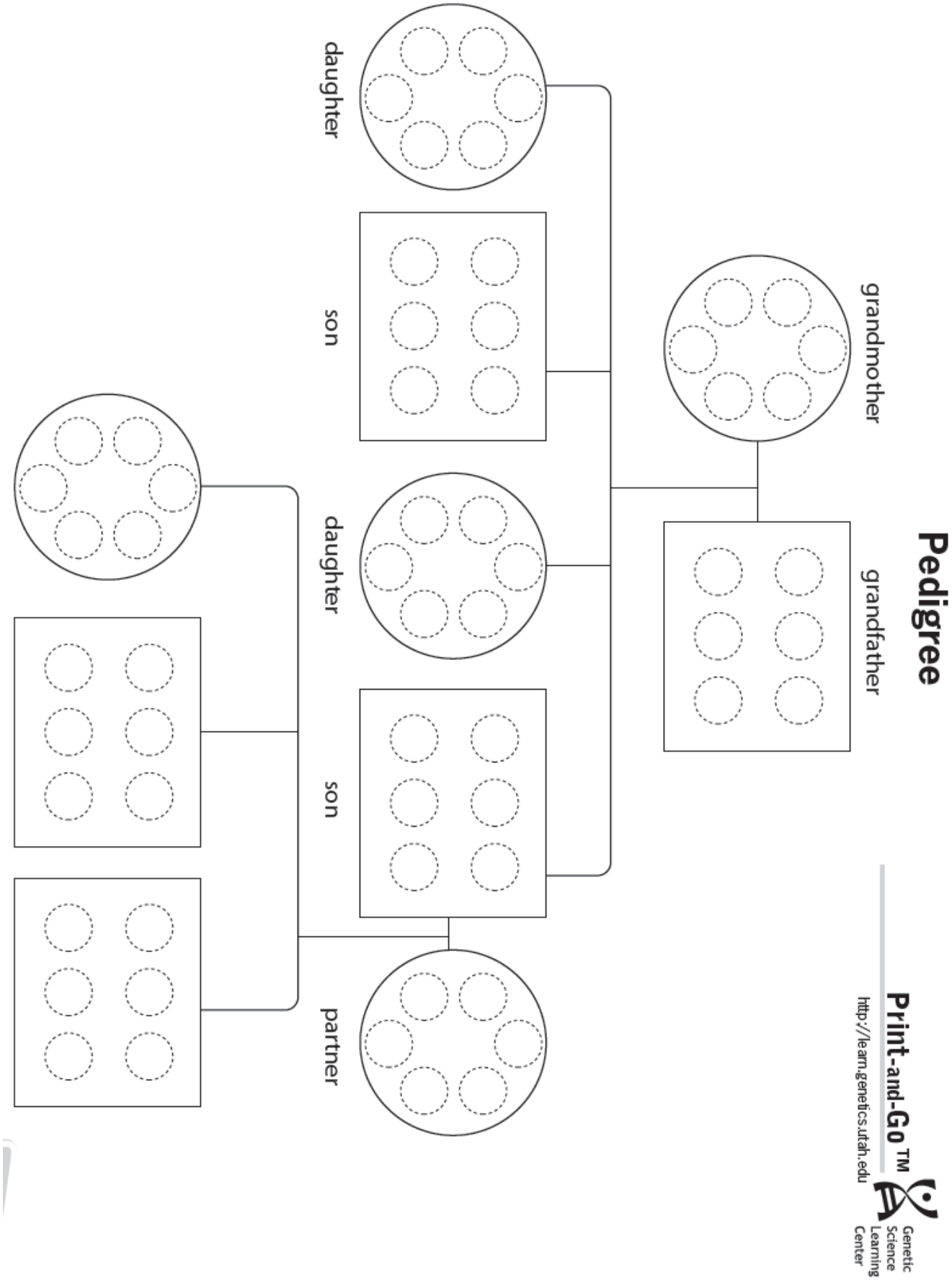
13. The couple has children. With closed eyes, mix and randomly draw out 3 pom poms from each parent (6 pom poms total). This will represent the genetic information inherited by their first child.

14. On the pedigree, color in the combination of pom poms that were passed on to the child.

15. Return the pom poms to the appropriate parent.

16. Repeat steps 12-14 to determine the genetic make-up of the other two children represented in the pedigree.

17. Label each individual that has been added to your pedigree as low, medium or high risk.


Pedigree
grandmother
grandfather
daughter
son
daughter
son
partner
Print-and-Go™
http://learn.genetics.utah.edu
Genetic Science Learning Center

Is it a boy or a girl?

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 (= $2n$).

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 (= $1n$). 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.

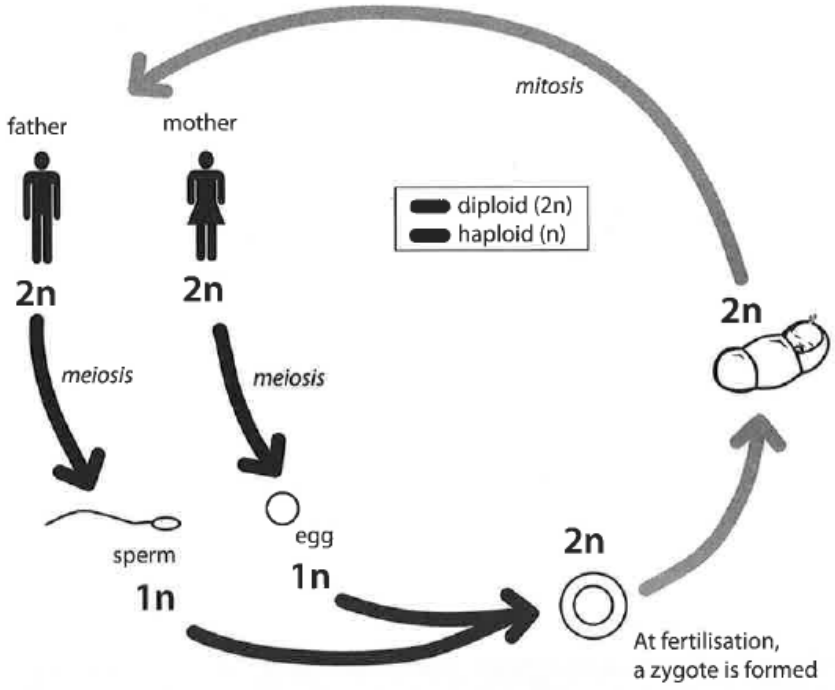


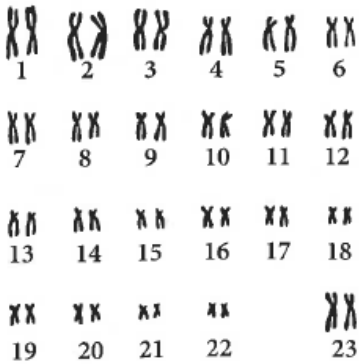
Figure 1

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

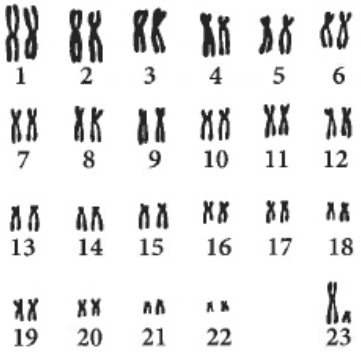
2 Find the terms 'diploid' and 'haploid' in figure 1. What do you think these terms mean?

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.

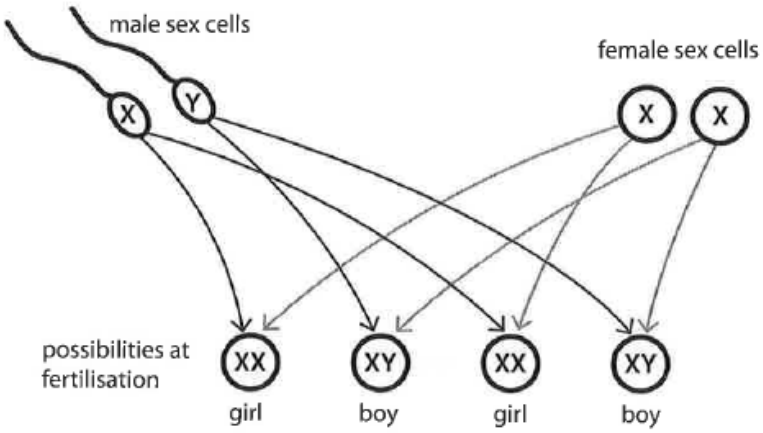


Figure 3

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?

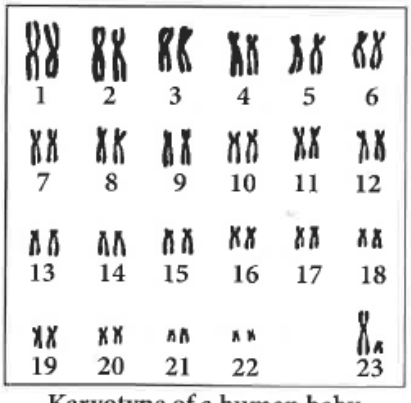
5 Describe how the sex of a human baby is determined.

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?

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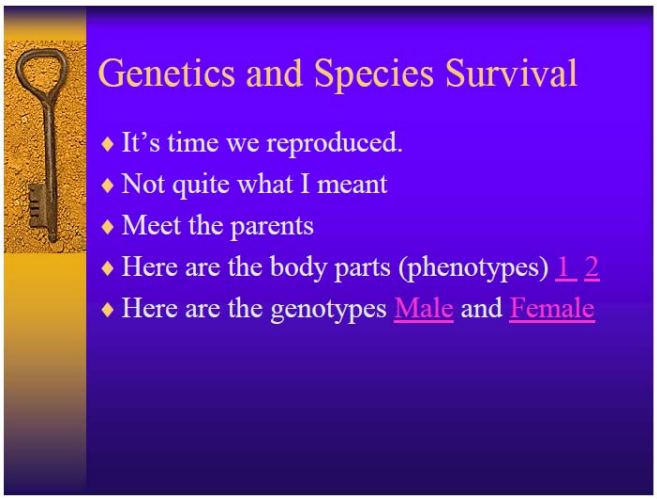
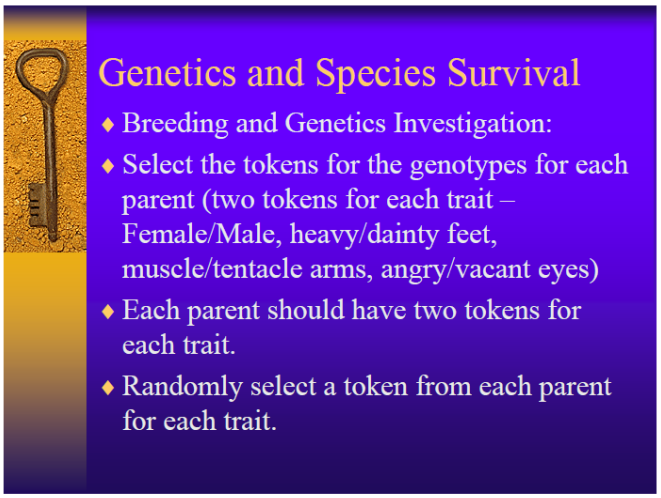
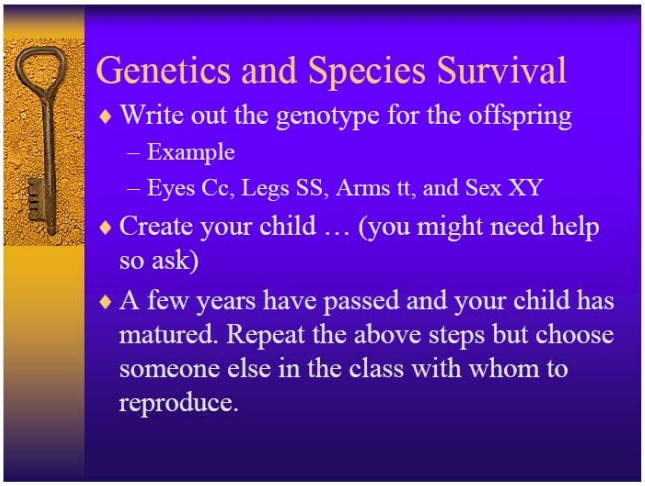
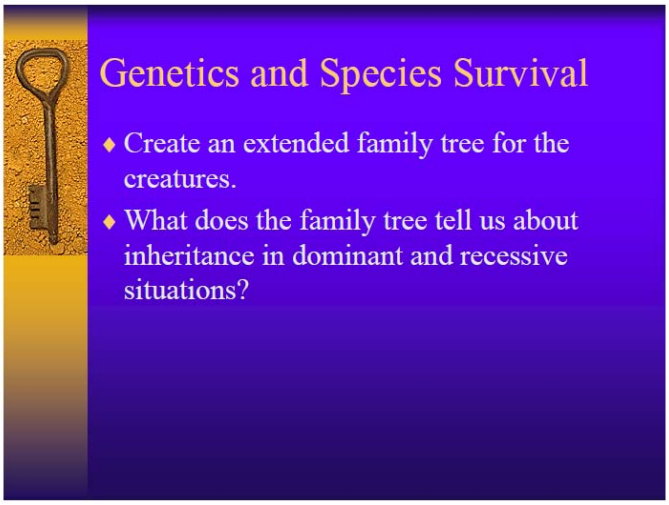








Sex Eyes Legs Arms

Male Genotype:

Female Genotype:

Offspring Genotype:

Grand Offspring Genotype:

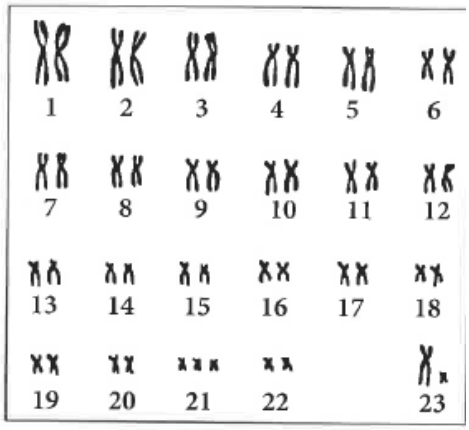
Genetic diseases – chromosomal

Genetic diseases occur when there is not the right number of chromosomes or when genes on chromosomes are not arranged correctly. Chromosomal disorders can occur when chromosomes fail to separate during the first meiotic division. As a result, one gamete may contain both chromosomes of one pair and the other has none. Deletions can also occur when a whole chromosome or part of a chromosome becomes lost. Radiation, some chemicals and excessive heat may cause mistakes to occur during meiosis.

The diseases described below are caused by chromosomal abnormalities.

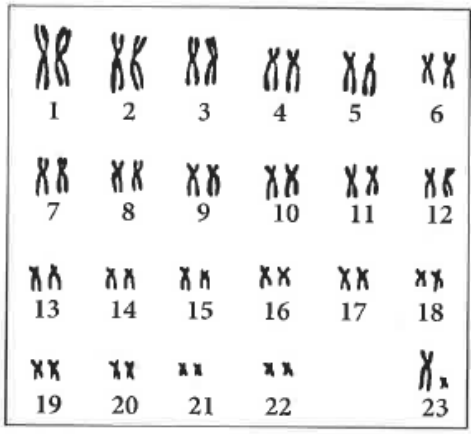
Down Syndrome

This disorder can be recognised by these characteristics: a short stature, stubby fingers, an eyelid fold, a wide gap between the first and second toes, a round head, a large tongue. The large tongue causes difficulties with speaking clearly. Another symptom is mental retardation. Down Syndrome is also called Trisomy 21, because there are three copies of the 21st chromosome, instead of two. The likelihood of a woman having a child with Down Syndrome increases rapidly with the age of the woman, from about 35 years. The frequency of Down Syndrome is about 1 in 800 births for women under 40 years of age and 1 in 80 for those over 40 years.



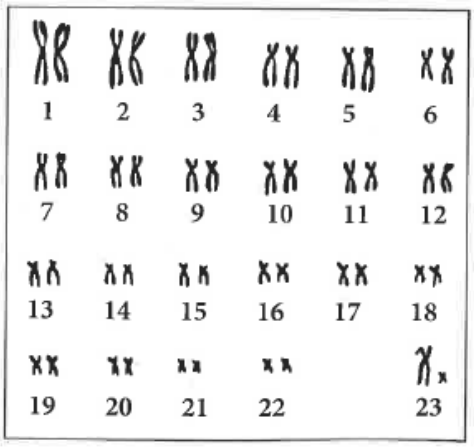
Cri du Chat Syndrome

A chromosome deletion is responsible for this disorder, which is also known as 'cat's cry syndrome'. Part of one of the 5th chromosomes is missing. An infant with this syndrome has a small head and a moon face. The baby's cry sounds like the meow of a cat, because of a malformed larynx. An older child has misshapen ears which are placed low on the head, and an eyelid fold. As the child matures, mental retardation becomes evident. Cri du Chat Syndrome occurs in about 1 of 50 000 births.



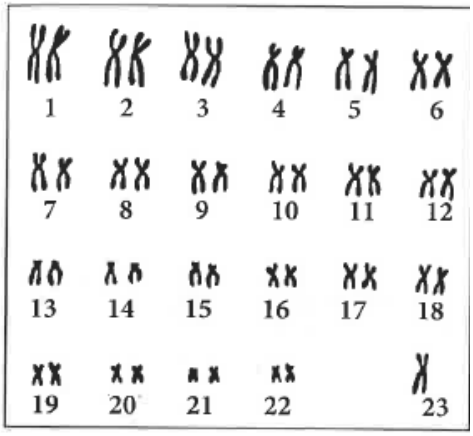
Fragile X Syndrome

With this condition, one X chromosome is nearly broken, leaving the tip hanging by a thread. Children with Fragile X syndrome appear to be normal, however they may be hyperactive or withdrawn. Their speech is delayed in development. When they become adults, they are short in stature and have a long face. Their jaw is prominent and they usually have protruding ears. Males with Fragile X Syndrome have large testicles, stubby hands and a heart defect. Fragile X Syndrome occurs in about 1 in 1 000 male births and 1 in 2 500 female births.



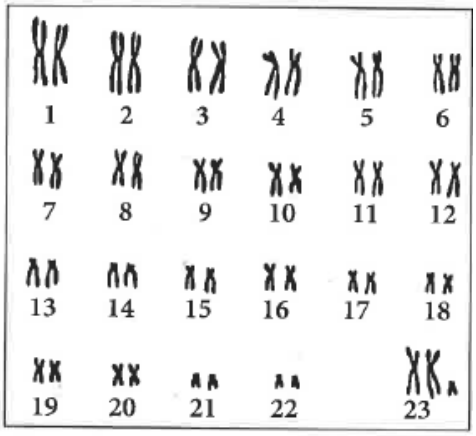
Turner Syndrome

With this condition, too few sex chromosomes have been inherited; the sex chromosomes are written as XO. The O signifies the absence of the second sex chromosome. Females are short and have a broad chest and webbed neck. Their ovaries, oviducts and uterus are very small and non-functional. They do not undergo puberty or menstruate and there is a lack of breast development. They are of normal intelligence and can lead normal lives. The frequency of Turner Syndrome is about 1 in 6 000 births.



Klinefelter Syndrome

These males have inherited two X chromosomes as well as the Y chromosome. As a result, they are sterile. Their testes and prostate glands are underdeveloped and they have no facial hair. They can also have some breast development. These individuals have large hands and feet as well as long arms and legs. They are slow learners but are not mentally retarded. Klinefelter Syndrome happens in about 1 in 1 500 births.



1 For each genetic disease, circle the chromosome responsible in the karyotype.

2 In your workbook, draw up a table like the one below. Complete the table to summarise the information about each inherited genetic disease.

Name	Cause	Symptoms	Frequency

3 Draw lines to match each genetic disease with the affected chromosomes.

a Cri du Chat Syndrome **b** Down Syndrome **c** Klinefelter Syndrome **d** Turner Syndrome

- deletion in chromosome 5
- XO
- XXY
- extra chromosome 21

Refer to the text below to answer the following questions.

Women over the age of 40 years are warned that they have a greater risk of delivering a baby with Down syndrome. These mothers are given the option to take a test to determine whether their baby has Down syndrome. Then they can make a decision whether to have the baby or not.

Some human rights groups argue that these tests are unethical. They say that every baby has a right to live regardless of whether they have disabilities or not. Other groups believe it is a mother's right to avoid the suffering and hardship with a disabled baby.

Be prepared to discuss your answers to these questions in class

4 Explain the meaning of:

a unethical. **b** right to live.

5 Explain why suffering and hardship may be experienced when a family has a child with Down Syndrome.

6 Do you think every pregnant woman over 40 years should have to take a Down Syndrome test?

7 **a** Do you think that parents should be allowed to use these tests to decide the future of their child?

b Do you think that doctors or welfare workers should be allowed to use these tests to decide the future of their patients' child?

Video evidence will be collected for these tasks.

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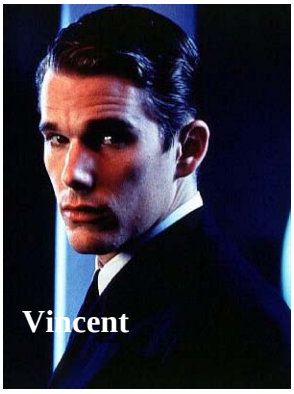
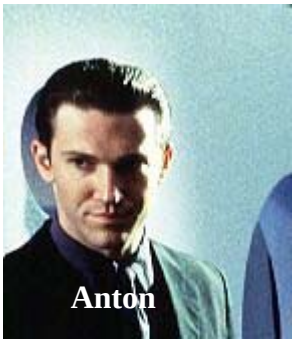
Gattaca





Compare and contrast the differences in the conceptions of Vincent and his younger brother Anton.

Vincent was treated differently than Anton. What were the reasons for this?

Why did Vincent leave home?

How do they choose people for careers in the future world shown in Gattaca?

What is the first information people want to find out about others? and how do they go about getting the information?

How did genetic techniques solve the crime?

Would it be correct to say that because of the police's reliance on genetic information the crime took longer to solve? Argue your case.

What are the advantages of knowing so much about a person's genetic information?

What are the disadvantages of knowing so much about a person's genetic information?

Would it be a good thing to use genetic information to decide the work that people should do? Argue your case.

Cloning animals – model discussion

1 The discussion text type on the previous page contains four sections: ***Introduction, Arguments for, Arguments against, Conclusion***. Draw a large square bracket in the left margin to indicate each section, and label it.

2 Often a paragraph includes key words near the start to describe what it is about. Double underline the key words in each paragraph.

3 In the right margin, briefly summarise the main content of each paragraph.

4 State the purpose of this discussion.

---

5 The arguments in a discussion are usually backed up with facts. Scientific words can highlight these. Circle at least five scientific words in the arguments section.

6 Although discussions contain many facts, they also use emotive language that exaggerates and appeals to your feelings. One example of emotive language is 'murdered'. Underline at least five other examples.

7 Select one topic from the list below and write a discussion about it. Start by planning useful points for each paragraph. You may need to research the topic first. In the scaffold at the bottom of the page, note useful points you could include for each paragraph. Then in your workbook write your discussion in six paragraphs.

Topics

a In the USA, people can pay $50 000 to have their favourite dead cat cloned. Is that wise?

b A farmer wants her ageing prize ram cloned so that she can continue to use it for breeding.

c Phar Lap was a very fast Australian racehorse and he died in 1932. He should be cloned so he can win horse races again.

d A cow producing unusually large amounts of milk is going to be cloned 12 times to replace a large but old cow herd.

e More mice should be cloned so that scientists can learn more about cloning techniques.

Discussion scaffold

1. Introduction	2. First argument for	3. Second argument for
4. First argument against	5. Second argument against	6. Conclusion and recommendation

Kinds of cloning

Read the text about cloning and then complete the task on the next page.

Explanation of cloning

A clone is an organism which is genetically identical to another organism. Cloning is a form of asexual reproduction which is widespread in nature. For unicellular organisms and many plants, it is a normal process, such as cell division or vegetative propagation. Mammals can produce identical twins when very young embryos split in two.

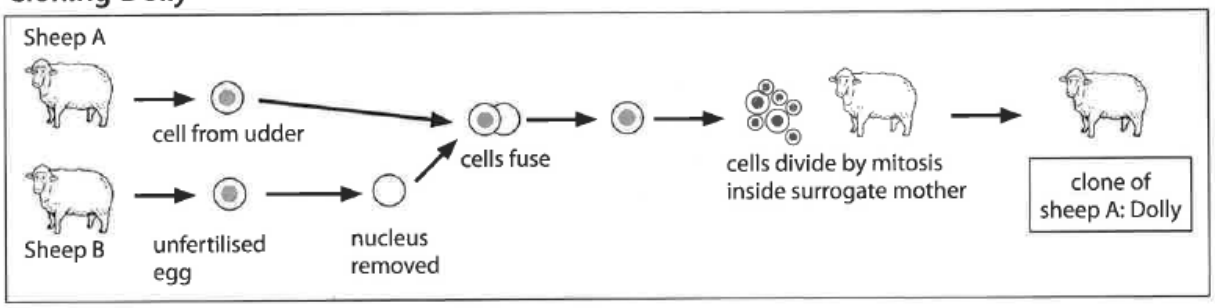
However, artificial cloning is the cause of much debate. There are two types of artificial cloning: embryo splitting and nuclear transfer.

- **Embryo splitting** is similar to the production of twins or multiple births, except it does not happen naturally – it is brought about by a scientist or doctor. In this type of cloning, the clones are the same age.
- During **nuclear transfer**, the complete set of chromosomes of one individual (the genetic parent) is transferred into a stem cell of another individual (the cell donor parent) and carried by a third individual (the host parent). This method allows the chromosomes to be transferred at different times and so the clones can be of completely different ages. For example, a pet cat that has died could be cloned for its owners.

It is also possible to transfer only part of a chromosome of an individual to a complete set of chromosomes of another individual. For example, a strawberry could carry a fish gene to prevent it from freezing at very low temperatures. This method produces **transgenic species**, which could not be produced in nature.

While embryo splitting has a fairly high success rate, nuclear transfer has often resulted in the death of the new clones or in new clones with health or development problems. Dolly, the cloned sheep, was one survivor of 300 clones. The diagram below outlines the process involved in producing Dolly the cloned sheep.

Cloning Dolly



Applications of cloning

In the process of gene farming, transgenic bacteria or animals are used to produce medicines. The advantages of medicines produced by biogenetic manufacturing processes are that they can be obtained in a much purer form than with conventional techniques that involve animal and human intermediate products. Production can be on a large scale and relatively cheap. However, there are also risks to the bacteria and animals due to their transgenic manipulation. People taking those medicines may not respond favourably and could become sick. Careful testing of drugs produced by gene farming is necessary.

In gene targeting or gene knock-out, individuals can be given favourable genes or have unfavourable genes destroyed. This would result in healthier offspring or offspring with certain characteristics. For example, cows could produce milk with a higher protein content. Using this process on animals would also help scientists learn how to cure genetic diseases in people.

During tissue cloning, particular human tissues are grown from the patient's cells rather than using donated tissue or animal tissue. The theory is that the patient's immune system would not reject the cloned tissue because it would not be made from foreign cells. For example, the burnt skin of a patient could be replaced with the patient's own but cloned skin.

Another idea for cloning human tissues involves swapping animal genes with human genes so that the animal grows a particular human organ. This organ would then be transplanted into the human.

Artificial insemination of farm animals, during which a female animal is fertilised with collected sperm from a particular male, has been occurring for a long time. However, it is a slow process. Usually only one valuable offspring is born at a time. If the chromosomes of a desirable animal could be used to grow clones, many more valuable offspring could be produced. This could result in great commercial and economic advantages. Even dead animals could be cloned.

Concerns about cloning

When cloning occurs in nature, more of already existing organisms are produced. This also happens during embryo splitting and complete nuclear transfer. However, artificial cloning is expensive and wasteful, although it could produce desirable and valuable organisms. As scientists practise cloning, they learn more about genetics, cloning techniques and development. Yet somebody must decide what is desirable and valuable, who should own those organisms, and what happens to the 'old' organisms.

Transgenic species are organisms that do not exist in nature; they are artificially made. It is not known what effect transgenic species could have on nature, if they were released. Once they are released, it may not be possible to capture them again. Potentially, they could interbreed and cause disease and destruction. Many questions about the possible consequences of cloning remain unanswered.

Task

Select one type of cloning and brainstorm possible applications and examples. You could use a mind-map to help organise your ideas. Then write a discussion:

- Start with an introduction in which you describe and explain your type of cloning.
- Develop two elaborated arguments which explain why that type of cloning should occur, and also develop two elaborated arguments against that type of cloning occurring.
- Finally, make a recommendation based on your arguments.

Don't forget to use persuasive language such as *should, must, always, never, only, rarely, extremely, under no circumstances* in order to convince the reader. Use conjunctions such as *on the other hand, however, as well as, but, since, when, because* to link your thoughts together.

To help you structure your discussion, use the scaffold below. Note useful points you could include for each paragraph. These notes will form the basis for your written discussion. Then in your workbook, write your discussion in six paragraphs.

Discussion scaffold

1. Introduction	**2. First argument for**	**3. Second argument for**
4. First argument against	**5. Second argument against**	**6. Conclusion and recommendation**

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