Mapping the Human Genome

Science content

Inheritance, genes, genetic diseases, DNA, bases, genetic engineering.

Science curriculum links
AT4 Genetics and evolution
AT17 The nature of science

Syllabus links
○ GCSE Science, Biology

Cross-curricular themes
○ Health Education

Lesson time
1–1½ hours
(including homework)

Links with other SATIS materials
309 Microbes Make Human Insulin
609 Hitting the Target
1204 From Babylon to Biotechnology

NERIS
Search on
DNA and UPPER SECONDARY
or on
GENES and UPPER SECONDARY

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SUMMARY

The unit describes the aims and complexity of the Human Genome Project, the costs and timescale and possible benefits to society. Students are asked to consider some of the project's moral implications.

STUDENT ACTIVITIES

☐ Reading, answering questions and making a glossary.

☐ Small group discussion of the moral implications.

AIMS

☐ To link with work on heredity

☐ To provide an understanding of an important scientific endeavour requiring unprecedented international cooperation

☐ To show students a developing area of science whose progress they may follow during subsequent years

☐ To provide a forum for discussion of the moral and ethical implications of this research

USING AND ADAPTING THE UNIT

☐ The unit draws together many of the concepts of genetics and is thus more appropriate for students towards the end of their GCSE work.

☐ The reading activities are suitable for independent study and may be done for homework.

☐ The discussion activities provide students with an opportunity to reflect on, share and evaluate their opinions. These questions may best be tackled in small groups with time afterwards for groups to share their thoughts with the class. The issues involved may impinge upon sensitive family matters and need sympathetic handling.

OTHER RESOURCES

☐ The progress of the Human Genome Project is often reported in the *New Scientist* magazine.

Answers to the questions

The information in brackets, [....], is not given in the text.

Q1 *Base, gene, chromosome, cell.*

Q2 *Finding the sequence of bases and positions of genes in the human genome.*

Q3 *3000 million bases*

(a) at 1980 prices (£5 per base) = £1.5 × 10^{10} or £15,000 million

(b) at 1990 prices (£1 per base) = £3 × 10^{9} or £3,000 million

(c) using automatic machines (£0.10 per base) = £3 × 10^{8} or £300 million

Q4 *Colds, sore throats and measles are diseases which you catch when infected by bacteria or viruses. Haemophilia and Huntingdon's chorea are hereditary diseases which are passed from parent to child in the genes.*

Q5 ***Genes*** *are responsible for inherited characteristics. They form parts of the DNA in cells. [Each gene is responsible for the production of one protein.]*

A defective gene *may give rise to a hereditary disease. [It is one which has an error in the groups of atoms, or in the sequence of bases of which it is composed.]*

Characteristics *are the features like eye colour, height, etc., which distinguish one individual from another.*

Chromosomes *contain the genes. [They can be seen when the cell divides. Humans have 46 chromosomes arranged in 23 pairs.]*

DNA *(deoxyribonucleic acid) is a polymer molecule [with two helical strands linked by the four base-pairs. Its structure allows it to make faithful copies of itself.]*

Human genome *is the sequence of genes that gives a human being its characteristics.*

Sequencing *is finding the order of the bases and also the location of each gene on the DNA.*

Mapping the human genome *– the result of sequencing: finding the detailed structure of human DNA.*

Moral implications *– distinguishing between the good and bad outcomes of this research.*

Discussion questions

Here are some possible answers.

1 **a** C; **b** B; **c** A; **d** B; **e** C; **f** C; **g** A.

2 Not much – the similarities between human genes will be much greater than any differences.

3 This is very much a matter of opinion. However, insurance companies might ask for the information as part of an insurance agreement (as they do regarding the risk of AIDS) or offer lower premiums to those who provide the information.

4 Personal opinions.

5 Personal opinions.

6
- How financial profits from this research may be distributed.
- Who should be allowed access to an individual's genome map.
- How society should decide and regulate the uses to which genome information is put.
- Whether individuals should have their genome sequenced at birth.
- Whether doctors should be able to withhold genome information from individuals.
- What developments will and will not become possible once the human genome has been mapped.
- How to prevent this information being used immorally – for example, breeding a submissive class of servants.

Acknowledgements

Sir Walter Bodmer, FRS, of The Imperial Cancer Research Fund read and commented on the draft of this unit.

Figure 3 by permission of The Lawrence Berkeley Laboratory, Human Genome Center.

Figures 2, 4 and on page 5 by Joyce Curtis.

Mapping the Human Genome

Project HUGO

This unit contains reading, questions and material for group discussion.

John is a haemophiliac. If he cuts himself, he will bleed to death unless he is given special treatment. He has haemophilia: his blood cannot clot.

John inherited haemophilia from his mother. She passed on a defective gene that stopped his blood clotting.

Imagine if doctors had known exactly what was wrong with the defective gene. Then they would have been had a much better chance of treating John's haemophilia successfully.

That's just one of the dreams of the scientists on Project HUGO – the Human Genome Project.

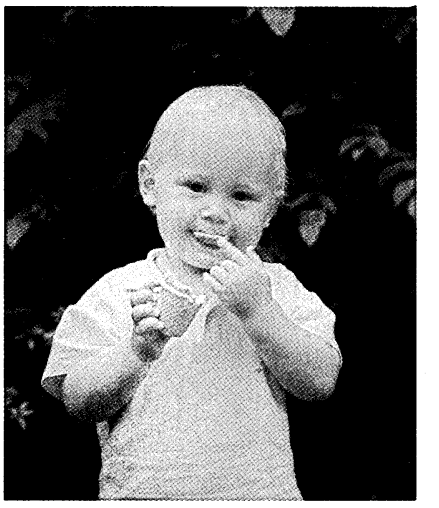
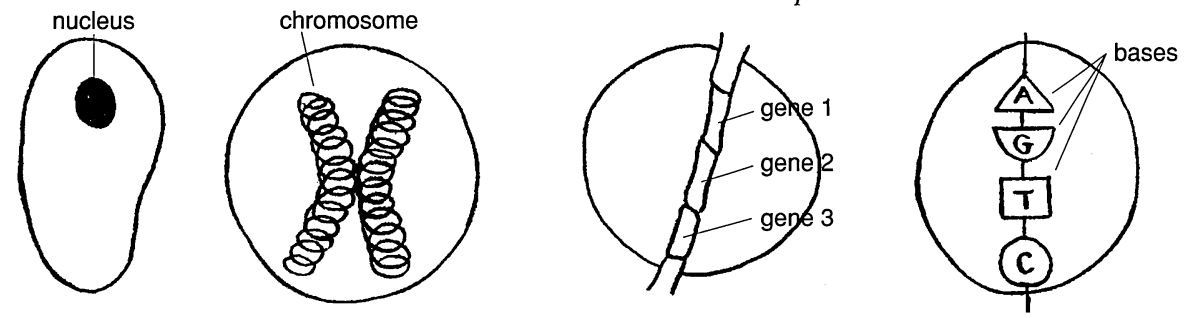


Figure 1 John suffers from haemophilia

What are they trying to do?

The goal of the HUGO project is to draw an exact map of all the genes that make up a human. Figure 2 shows what this means.

Figure 2



The nuclei of cells contain ...

chromosomes which contain ...

... genes, which are made up of ...

...DNA, which has millions of bases joined in a particular order.

You get your characteristics – eye colour, height, features and so on – from your parents. You inherit **chromosomes**; 23 from your mother and 23 from your father. The chromosomes carry genes, and each gene decides a particular characteristic. So there are genes for eye colour, hair colour and so on. You have your own set of up to 100 000 genes with copies in every cell.

Genes and chromosomes are made of a chemical called **DNA** (deoxyribonucleic acid). DNA is a polymer, and it's made of parts called nucleotides which carry four different **bases** joined together in a particular order. The bases make up a code – the genetic code which decides your characteristics.

Q1 *Arrange the following in order of size:*
a gene,
a base,
a human chromosome,
a human cell.

If you joined together all the genes in a human being end to end you would make a string of 3000 million bases. This is the **human genome**. It is so long that if you printed it the result would fill 200 large telephone books. The molecule itself would be two metres long if you stretched it out.

At the moment we can't print out all the bases in the right order. That's what HUGO is about – *to find the exact order, or sequence, of all the bases that make up the human genome*. Finding out the order of bases is called **sequencing**. But it doesn't simply mean finding the sequence of bases. It also means finding where each gene is, and the sequence of bases in each gene.

It's like finding the recipe for a complete human being.

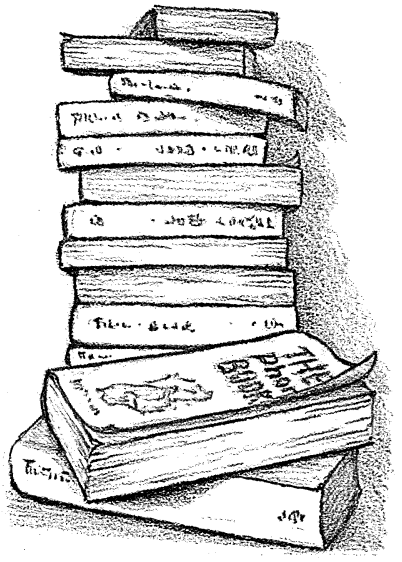
How are they doing it?

Scientists already know how to map parts of the human genome. In fact, some of the work has been done already. Several million bases of human DNA have been mapped – but that still leaves 99.9 per cent to do! If scientists continued working on the problem at this rate, it would take them 500 years.

However, the aim of HUGO is to complete the map by the year 2005. That means an enormous research effort by thousands of scientists. HUGO is too big to be done in a single laboratory. It will involve dozens of laboratories all around the world.

Basically, the method is to chop up the DNA into short pieces using special enzymes. Then the bits are pieced together to make the full sequence, like doing an enormous jigsaw puzzle. The problem is, the jigsaw has several thousand million pieces! Just keeping track of it all is a huge job, involving enormous computer systems (figure 3).

The scientists will spend much of the first five years developing their sequencing technology and finding out where each gene is. They will practise on simpler organisms like yeast, worms and mice. As they improve their skills and technology they will become quicker and more accurate at sequencing DNA.



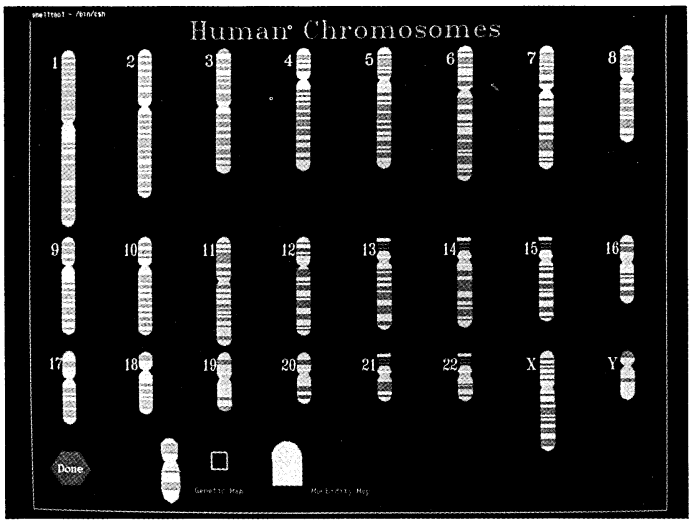
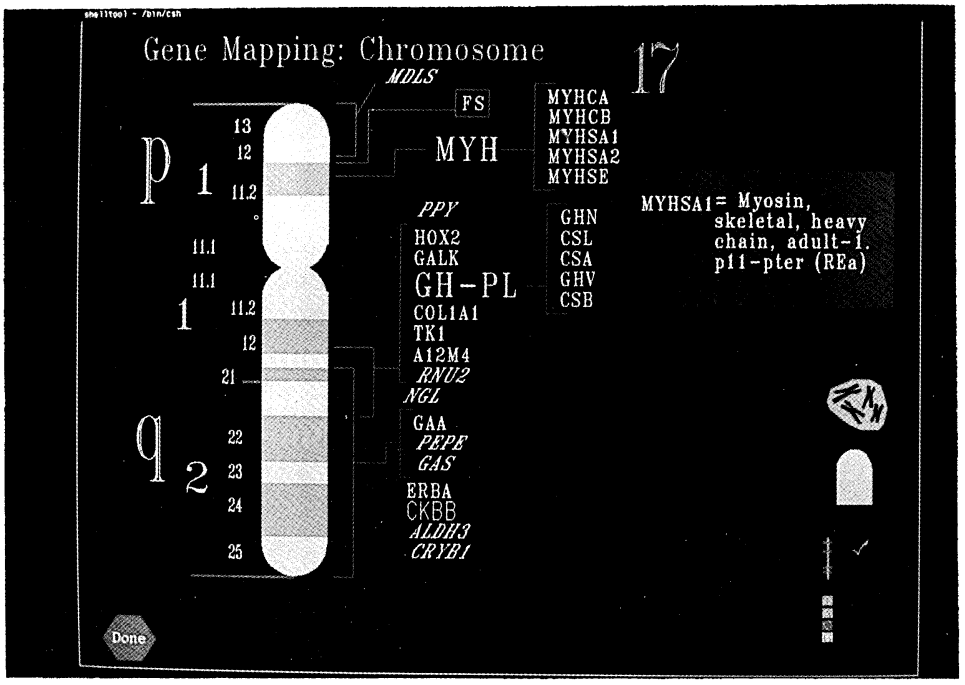
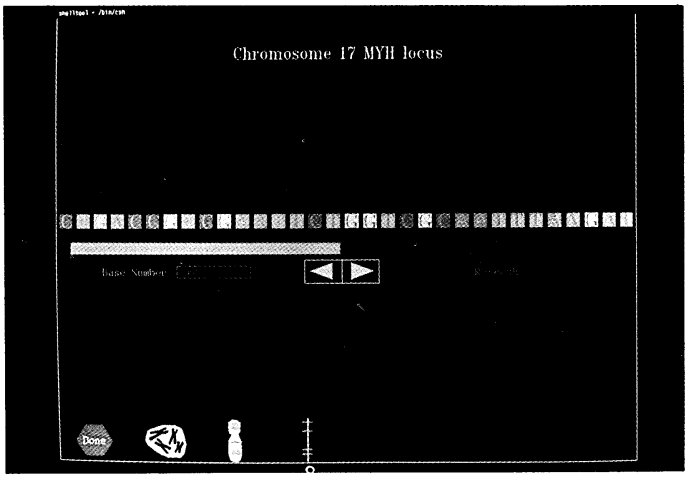


Figure 3 Scientists working on the human genome project are developing computer workstations so that they can share information.





The pictures show how a scientist is able to home in on a particular chromosome.

And the cost?

The project will be enormously expensive. In 1980 the cost of finding the sequence of a fragment of DNA was about £5 per base. So a fragment of DNA 100 bases long cost £500 to sequence. There are 3000 million bases in the human genome. You can see it's an expensive business!

Q2 What is Project HUGO is trying to achieve?

Q3 What would be the cost of mapping the human genome
(a) at 1980 prices,
(b) at 1990 prices,
(c) using automatic machines?

Fortunately, as scientists do more of this work, they get better at it. They can do it quicker – and more cheaply. In 1990, the cost was under £1 per base – and scientists are developing automatic machines that can do it for about £0.10 per base.

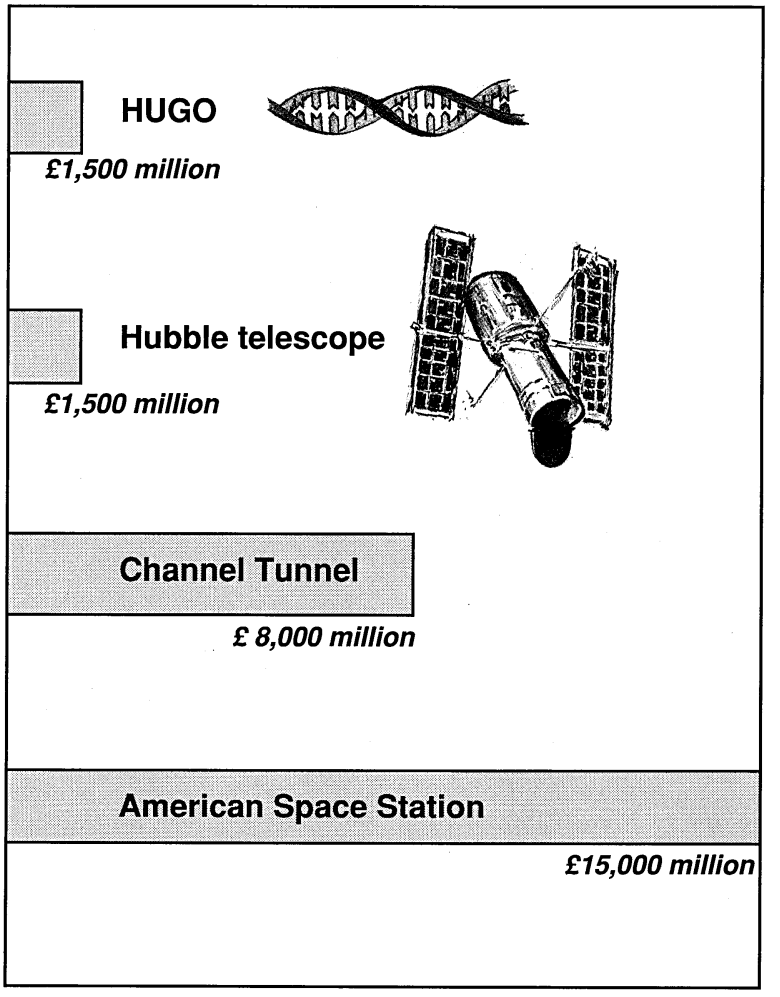


Figure 4 The costs of large research projects for comparison

The HUGO project is expected to cost about £1,500 million altogether. Figure 4 shows how this compares with the cost of other big projects. It sounds expensive, but it's actually only about one twentieth of the annual cost of the National Health Service.

What does the money buy?

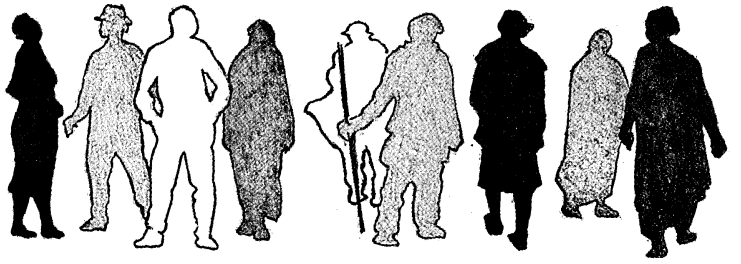
A map of the human genome could be the key to curing hundreds of genetic diseases like haemophilia and Huntingdon's chorea – where the patient suffers mental deterioration and early death.

Many common diseases like heart disease, mental disease and arthritis have a genetic link, so that people who inherit a particular gene may be more likely to suffer from the disease. Mapping the genome would make it possible to investigate this. And most scientists agree that the key to cancer is an understanding of the human genome.

And there are many other possibilities. What if parents who were short wanted a tall child? What if parents wanted to produce a child that would become the fastest sprinter or the greatest leader? If scientists knew what all the genes did, they might be able to grant parent their wishes . . . at a price.

Some time in the future, it might be possible for everybody to have their own genome mapped at birth – or before. This would be a plan for a copy of that person. Think of the implications of *that*. You will be able to think of many other possibilities – and you can discuss them later.

The scientists who are organising the human genome project are very concerned about its moral implications. They have decided to set up groups to discuss the issues, such as the way genetic information about people could be misused.



The moral implications

Why are people concerned about the moral implications of mapping the human genome?

Once you have mapped the genome, you have the complete plan for a person. You could change it . . . . You might be able to design human beings to order.

That possibility is a long way off. But right now there are tricky problems to sort out. For example, suppose doctors discover that a patient has a gene which makes it more likely he will get heart disease. Who should be given the information? You can see there are some tricky things to decide.

Q4 *From what you know of diseases like colds, sore throats and measles, explain how they are different from diseases mentioned in the unit – haemophilia and Huntingdon's chorea.*

Q5 *New ideas and discoveries need new words to describe them.*
Look at how these words and phrases are used in the unit

- *gene*
- *defective gene*
- *characteristics*
- *chromosomes*
- *DNA*
- *human genome*
- *sequencing*
- *mapping the human genome*
- *moral implications.*

Make a glossary of the terms above – that is an alphabetical list of words or phrases with their definitions or explanations. You could add more scientific words used in the unit.

(You may find it useful to use a dictionary or any reference books you may have to hand.)

Organising the discussion

- Work in a small group.
- Appoint someone to chair the group and to report back to the class if required.

Some questions to discuss

1 Here is a list of suggestions for things that *might* become possible if we had a full map of the human genome.

- **a** Making people live forever
- **b** Making sure your children are born intelligent
- **c** Curing genetic diseases like haemophilia
- **d** Curing AIDS
- **e** Making it possible for humans to fly (without help)
- **f** Making it possible to live without food
- **g** Finding out if a person is particularly likely to develop arthritis

Say whether each one

- ***A*** *almost certainly could be done,*
- ***B*** *possibly could be done,*
- ***C*** *definitely could not be done.*

2 Would it matter which particular person's genes the scientists chose to map for the HUGO project?

3 Suppose doctors discover that a patient has a gene that makes heart disease more likely. Who should be told?

- a The patient.
- b The patient's close relatives.
- c The patient's employer.
- d The patient's insurance company.

4 Suppose that, some time in the future, you are the parent of a new baby. And that by then every individual can have their own genome mapped at birth.

(a) Would you have your child's genome mapped?

(b) Who should be allowed to see and use that map? What might happen if it got into the wrong hands?

5 If genome mapping becomes available, would you want to see your partner's genome map before you planned to have a child?

6 Scientists are investigating the moral implications of mapping the human genome. List five questions you would want them to consider.

Answers to the questions are given in the *Teacher's Notes.*