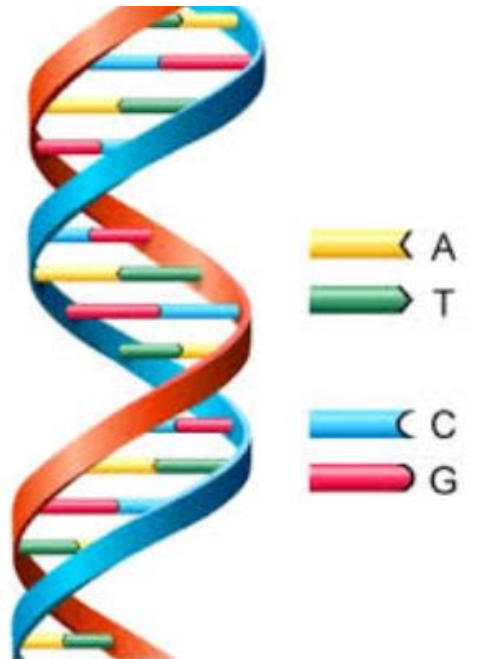


The Human Genome Project

Wednesday, 28 April 2021

Starter: Why is it important that we know about all the human genes?



The big picture: "How has the Human Genome Project impacted on human life?"

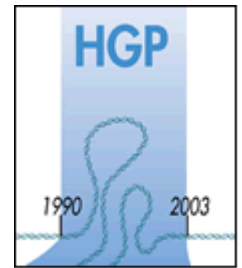
Learning outcomes

- ✓ Explain the aims of the human genome project.
- ✓ Analyse how the outcomes of the human genome project have been used to develop new drugs.
- ✓ Evaluate the ethical implications of the human genome project.

Genome

- All the DNA of an organism.
- It includes all the genes that carry all the genetic information to making all the proteins required by the organism

The Human Genome Project



- Started in 1986 (USA and UK) but officially 'started' in 1990, Europe and Japan joined in 1992
- Completed in 2003 - under budget and 2 years early
- The sequence is not that of one person, but is a composite derived from several individuals. Therefore, it is a "representative" or generic sequence. To ensure anonymity of the DNA donors, more blood samples (nearly 100) were collected from volunteers than were used, and no names were attached to the samples that were analysed. Thus, not even the donors knew whether their samples were actually used.
- Also sequenced yeast and animals used in medical research e.g. zebra fish and rats.

What were the aims of the human genome project?

- *To identify all the approximately 20,000-25,000 genes in human DNA.*
- *To find where each gene is located*
- *To determine the sequences of the 3 billion chemical base pairs that make up human DNA.*
- *Store this information in databases.*

Estimated time 15 years.

(started in 1980)

Estimated cost US\$3 billion



He used to work 80 hours a week sequencing genes for the Human Genome Project

Exam question:

Explain the aims of the human genome (4 marks)

Extra challenge: Why do you think scientists also sequenced the genomes of animals used for medical research?

LO: Explain the aims of the human genome project.

Self-assessment: Model answer

- To identify all the genes in the human genome.
- To find the location of all the genes.
- To determine the sequences of the base pairs that make up the human DNA.
- To find the functions of different genes.
- To publish the results on a public database.

Outcomes of the HGP

Using page 137-138 and the human genome website:

<http://www.genome.gov/25019879>

Fill in your table about the outcomes of the human genome project

LO: Analyse how the outcomes of the human genome project have been used to develop new drugs.

Outcomes of the HGP; Summary

Detailed information about the genome	<ul style="list-style-type: none">▪ 30 000 - 40 000 genes▪ average human gene contains 3000 bases▪ non-coding sequences (junk DNA) makes of 50%▪ 1.4 millions locations of single nucleotide polymorphisms
Identification of new genes	<ul style="list-style-type: none">▪ breast cancer gene▪ total colour blindness gene▪ genes analysed for mutations causing disease
Identification of new drug targets	<ul style="list-style-type: none">▪ a molecule that a drug interacts with▪ identification of genes allows identification of drug targets

<p>Preventative medicine and improved drug treatment</p>	<ul style="list-style-type: none">▪variation in base sequences may account for why some people experience side effects from drug therapies▪identification of mutations associated with a particular disease allows patient to make lifestyle changes or adopt preventative drug therapy
<p>Understanding basic biology</p>	<ul style="list-style-type: none">▪receptor proteins in the sense of taste▪post-production processing of proteins
<p>Investigating evolution</p>	<ul style="list-style-type: none">▪repeat sequences replicate and insert themselves into the DNA modifying, reshuffling and creating new genes▪comparisons with the genome of other organisms establishes evolutionary pathways

Ethical issues

List the key ethical issues raised by the Human genome project.

Potential disadvantages or ethical objections posed by the HGP

- People may be put under undue pressure to not have children or terminate pregnancies
- Increases pressure for germ line therapy to prevent children inheriting genetic conditions
- Embryo has no choice/say in the matter
- May lead to discrimination with jobs
- May lead to serious issues with insurance (life/medical/car etc.)
- May lead to 'designer babies' with selection for specific fashionable/on a whim characteristics

Potential disadvantages or ethical objections posed by the HGP

- Knowing something might happen may cause psychological stress/ some may not want to know (easier to cope if you don't know in advance?)
- Human rights/personal freedom: intrusion, infringement of civil liberties; who decides who should have genetic tests? Who decides who has access to/should have potentially expensive treatment
- Data protection issues: who has access to your genetic information?

Plenary

Past paper questions:

- 1) The HGP is making it possible to identify people who may be at risk of developing medical conditions such as heart disease, cancer or diabetes. Reasons why identifying people at risk might be of benefit to the people who are tested. (2 marks)
- 2) Suggest two reasons why carriers of breast cancer genes should not face discrimination when applying for jobs or insurance. (2 marks)

Check your answers

- 1) To warn people at risk to make life style changes, e.g. quit smoking, eat less saturated fat, do more exercise
 - Gives people a more informed choice in how to plan their lives
 - To plan medical provision/monitoring for the individual
 - To target drug treatment to delay onset/reduce symptoms
 - To determine healthcare priorities

- 2) It is not their fault that they have the gene
 - Most of us would prefer a society where such economic risks were spread across all of us
 - Such discrimination would reduce the encumber o people going for testing, which could be fatal for them
 - Having the gene does not mean you will necessarily develop the disease.

Resources

Outcomes	Explanation
Detailed information about the genome	
Identification of new genes	
Identification of new drug targets	
Preventative medicine and improved drug treatment	
Understanding basic biology	
Investigating evolution	