

19.2 Genetic technology applied to medicine

Name: _____ Class: _____ Date: _____ Total: 25 marks

KEY WORDS breast cancer • gene therapy • genetic screening
• insulin • recombinant human proteins

Objective

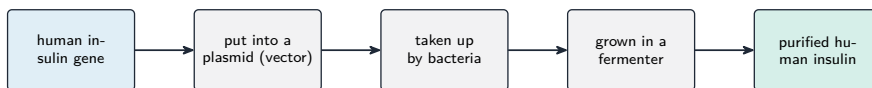
Build the skills to answer exam questions on **genetic technology applied to medicine** – **recombinant human proteins**, **genetic screening**, **gene therapy**, and their ethical issues.

You must be able to:

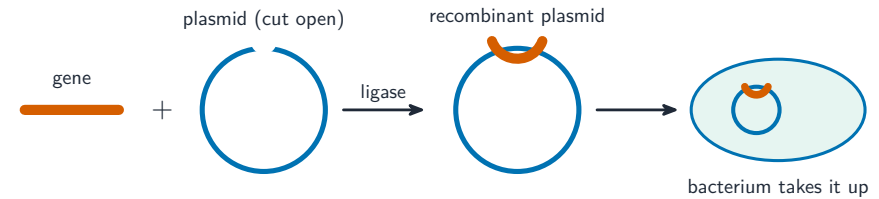
- explain the advantages of recombinant human proteins (insulin, factor VIII, ADA)
- outline genetic screening (BRCA, Huntington's, cystic fibrosis) and gene therapy (SCID)
- discuss social and ethical considerations

1 Key knowledge and worked examples

■ Recombinant human proteins



The insulin gene goes into bacteria, which become living factories – an exact human copy, never in short supply



A plasmid vector carries the human gene into bacteria

A human gene is placed in bacteria so they make a **recombinant** human protein – an exact copy. Advantages: it is identical to the human protein, never in short supply, and avoids using animal/donor proteins (so fewer allergic reactions or infections). Examples: **insulin** (diabetes), **factor VIII** (haemophilia), **adenosine deaminase** (a faulty immune system).

■ Genetic screening and gene therapy

Genetic screening tests DNA for disease alleles before symptoms appear – e.g. **BRCA1/BRCA2** (breast cancer risk), **Huntington's disease**, **cystic fibrosis**. **Gene therapy** puts a working gene into a patient's cells – used for **SCID** (failed immune system) and some inherited eye diseases.

■ Ethics

Concerns: who may see your results; misuse by insurers/employers; safety and permanence of changes; who decides. These are weighed against the benefits.

2 Practice

2.1 State **two** advantages of using recombinant human insulin rather than insulin from animals. [2]

2.2 Name **one** disease that can be detected by genetic screening. [1]

3 Exam-style questions

3.1 Gene therapy treats a genetic disease by: [1]

- ☐ **A** giving the patient a recombinant protein
B putting a working copy of a gene into the patient's cells
C screening the patient's DNA
D removing the faulty allele from all body cells permanently

3.2 Which is an advantage of making insulin from genetically engineered bacteria? [1]

- ☐ **A** it is slightly different from human insulin
B it is identical to human insulin and never in short supply
C it must be extracted from animal pancreases
D it cannot be purified

3.3 (a) Explain **one** advantage of genetic screening for a person with a family history of Huntington's disease. [2]

(b) Discuss **one** ethical concern raised by genetic screening. [2]

3.4 Explain how gene therapy could help a child with SCID. [2]

3.5 Human insulin for treating diabetes is produced by genetically modified bacteria.

(a) Describe how the human insulin gene is obtained and inserted into a bacterium, naming the enzymes and the vector used. [5]

(b) Explain why a marker gene is included, and describe how it is used. [3]

(c) Explain **two** advantages of using recombinant human insulin rather than insulin extracted from animals. [2]

(d) Distinguish between genetic screening and gene therapy, and state one ethical concern raised by each. [4]

Solutions

Each answer shows the steps, then the **result**.

2.1

- any two: identical to human insulin; never in short supply; fewer allergic reactions; no risk of animal-borne infection; no ethical objection from using animals

2.2

- any one: breast cancer (BRCA1/BRCA2); Huntington's disease; cystic fibrosis

3.1 B

- Gene therapy adds a working copy of the gene to the patient's cells

3.2 B

- Engineered bacteria make insulin identical to human insulin, in unlimited supply

3.3

(a)

- it lets the person know their risk before symptoms appear, so they can make informed choices about treatment, monitoring or having children

(b)

- e.g. results could be misused by insurers/employers, or cause anxiety / there is no cure for Huntington's

3.4

- a working copy of the faulty gene is inserted into the child's cells; the cells then make the missing protein, restoring immune function

3.5

(a)

- The mRNA for insulin is taken from human pancreatic cells and **reverse transcriptase** is used to make a complementary DNA copy of the gene; alternatively the gene is cut from the genome with a **restriction endonuclease**, which leaves sticky ends; a **plasmid** vector is cut with the same enzyme so its sticky ends are complementary; the gene and plasmid are joined by **DNA ligase**; the recombinant plasmid is then taken up by the bacterium, for example after treatment with calcium ions and heat shock

(b)

- Only a small proportion of bacteria take up the plasmid, so the transformed cells must be identified. A marker gene – for antibiotic resistance or for a fluorescent protein – is carried on the same plasmid; growing the culture on a medium containing that antibiotic, or under UV light, leaves only the transformed bacteria visible or alive

(c)

- It is identical to human insulin, so it works exactly as the body's own does and is far less likely to cause an allergic reaction; it can be produced in unlimited quantity by fermentation, and is acceptable to people whose religion or beliefs forbid animal products

(d)

- **Genetic screening** tests a person, embryo or fetus for alleles causing a genetic condition – it identifies risk but changes nothing; a concern is that the results could be used by insurers or employers to discriminate, or could pressure a couple's reproductive decision. **Gene therapy** inserts a working allele into a patient's cells to treat the condition; somatic therapy affects only that patient, but germ-line therapy would change every future generation, with consequences that cannot be predicted or consented to