A-LEVEL BIOLOGY • EXERCISE SHEET

# 19.2 Genetic technology applied to medicine

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

**KEY WORDS** breast cancer 乳腺癌 • gene therapy 基因治疗 • genetic screening 基因筛查 • insulin 胰岛素 • recombinant human proteins 重组人蛋白

## Objective

Build the skills to answer exam questions on **genetic technology applied to medicine** — **recombinant human proteins** 重组人蛋白, **genetic screening** 基因筛查, **gene therapy** 基因治疗, and their ethical issues.

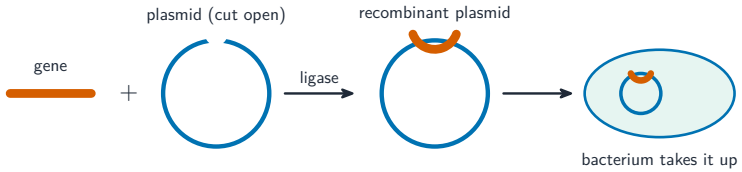
**You must be able to:**

- 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 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** 胰岛素 (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** 囊性纤维化. **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

**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.