

**OXFORD CAMBRIDGE AND RSA EXAMINATIONS
A2 GCE
F225/01**

HUMAN BIOLOGY

Genetics, Control and Ageing

THURSDAY 23 JUNE 2016: Morning

DURATION: 2 hours

plus your additional time allowance

MODIFIED ENLARGED 24pt

Candidate forename		Candidate surname	
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Centre number						Candidate number				
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Candidates answer on the Question Paper.

OCR SUPPLIED MATERIALS:

None

OTHER MATERIALS REQUIRED:

Electronic calculator

Ruler (cm/mm)

READ INSTRUCTIONS OVERLEAF



INSTRUCTIONS TO CANDIDATES

Write your name, centre number and candidate number in the boxes on the first page. Please write clearly and in capital letters.

Use black ink. HB pencil may be used for graphs and diagrams only.

Answer ALL the questions.

Read each question carefully. Make sure you know what you have to do before starting your answer.

Write your answer to each question in the space provided. If additional space is required, you should use the lined page(s) at the end of this booklet. The question number(s) must be clearly shown.

INFORMATION FOR CANDIDATES

The number of marks is given in brackets [] at the end of each question or part question.

The total number of marks for this paper is 100.



Where you see this icon you will be awarded marks for the quality of written communication in your answer.

You may use an electronic calculator.

You are advised to show all the steps in any calculations.

Any blank pages are indicated.

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Answer ALL the questions.

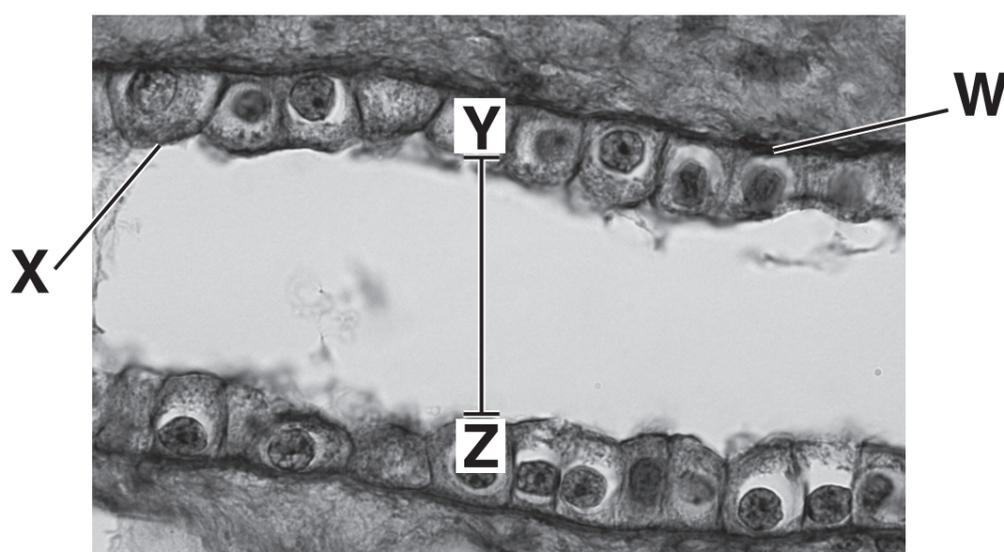
1 Excretion is the removal of metabolic waste products from the body. The kidney is one of the organs involved in excretion. One of the functions of the kidney is to remove nitrogenous waste.

(a) Name ONE nitrogenous waste product that is removed by the kidney.

_____ [1]

(b) Fig. 1.1 is a photomicrograph of a longitudinal section through the cortex of a kidney, showing the epithelial tissue lining a COLLECTING DUCT.

Fig. 1.1



magnification $\times 300$

- (i) Using the information in Fig. 1.1, calculate the diameter of the lumen of the collecting duct between positions Y and Z.

Show your working. Give your answer to the nearest whole number.

Answer = _____ [2]

- (ii) Suggest why the diameters of collecting ducts in the medulla of the kidney can be much wider than the diameters of collecting ducts in the cortex.

_____ [1]

- (c) (i) Name the hormone that binds to receptors at location W in Fig. 1.1, AND name the endocrine gland from which this hormone is released.

Hormone _____
Endocrine gland _____ [1]

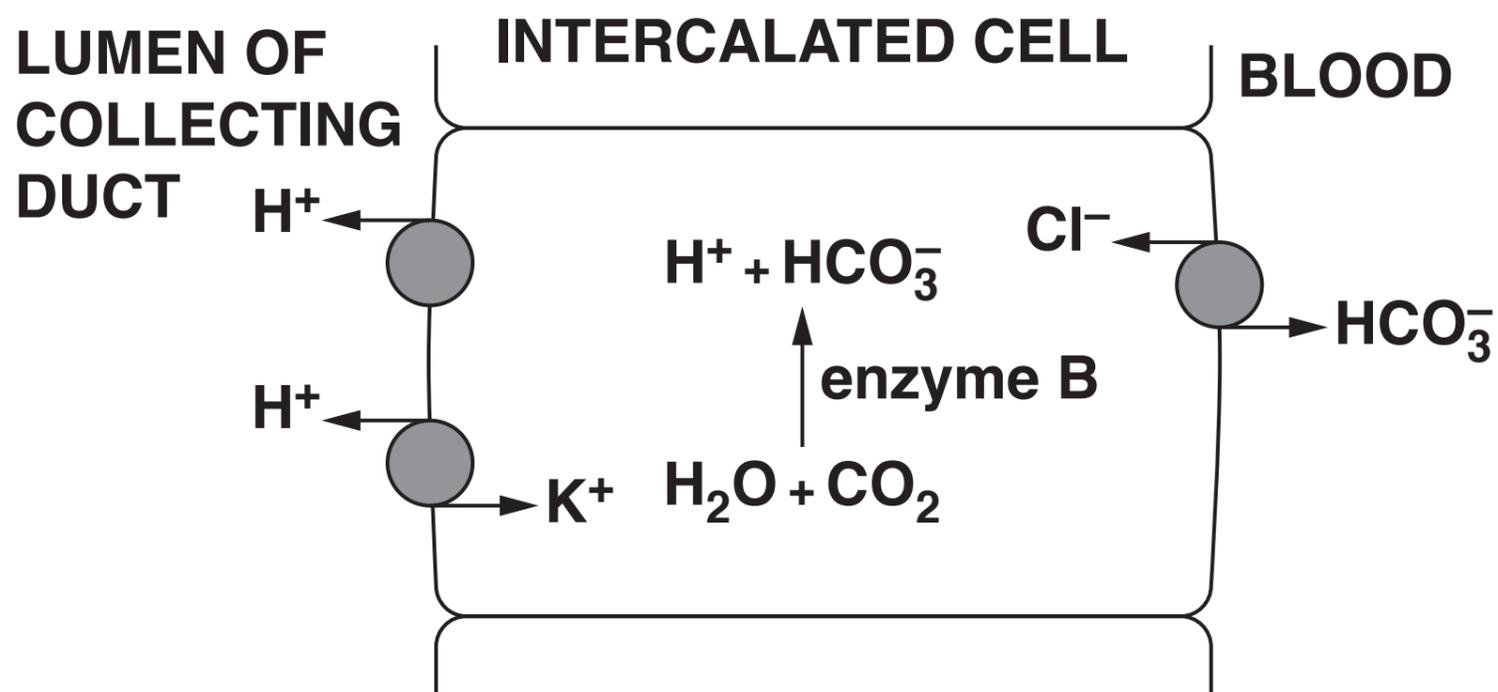
- (ii) Describe how the APPEARANCE of membrane X in Fig. 1.1 would differ from that of a cell lining the wall of a PROXIMAL CONVOLUTED TUBULE.

_____ [1]

- (d) Intercalated cells are specialised cells located in the walls of the collecting ducts. One function of intercalated cells is the regulation of the pH of blood plasma.

Fig. 1.2 is a diagram of an intercalated cell showing some of the mechanisms involved in the regulation of pH by the kidney.

Fig. 1.2



- (i) Name the enzyme labelled B in Fig. 1.2 that catalyses the reaction shown.

_____ [1]

- (ii) Using the information in Fig. 1.2, suggest why intercalated cells contain large numbers of mitochondria.

_____ [1]

- (iii) Identify ONE OTHER organ system in the body that is responsible for controlling the pH of blood.

_____ [1]

(e) The solute concentration of the fluid in the lumen of the collecting duct can vary.

If a health professional suspects that a patient may have diabetes insipidus, a water deprivation test may be carried out.

Part of the procedure for carrying out a water deprivation test is given below:

the patient does not drink for the duration of the test

the patient is weighed at the beginning of the test and after collecting each urine sample

urine samples are collected at regular intervals over a period of up to 9 hours

the solute concentration of each urine sample is measured.

(i) State ONE way in which the composition of urine of a person with diabetes insipidus will be different from that of a person with diabetes mellitus.

_____ [1]

(ii) Suggest why it is important to monitor the weight of the patient during the test.

_____ [1]

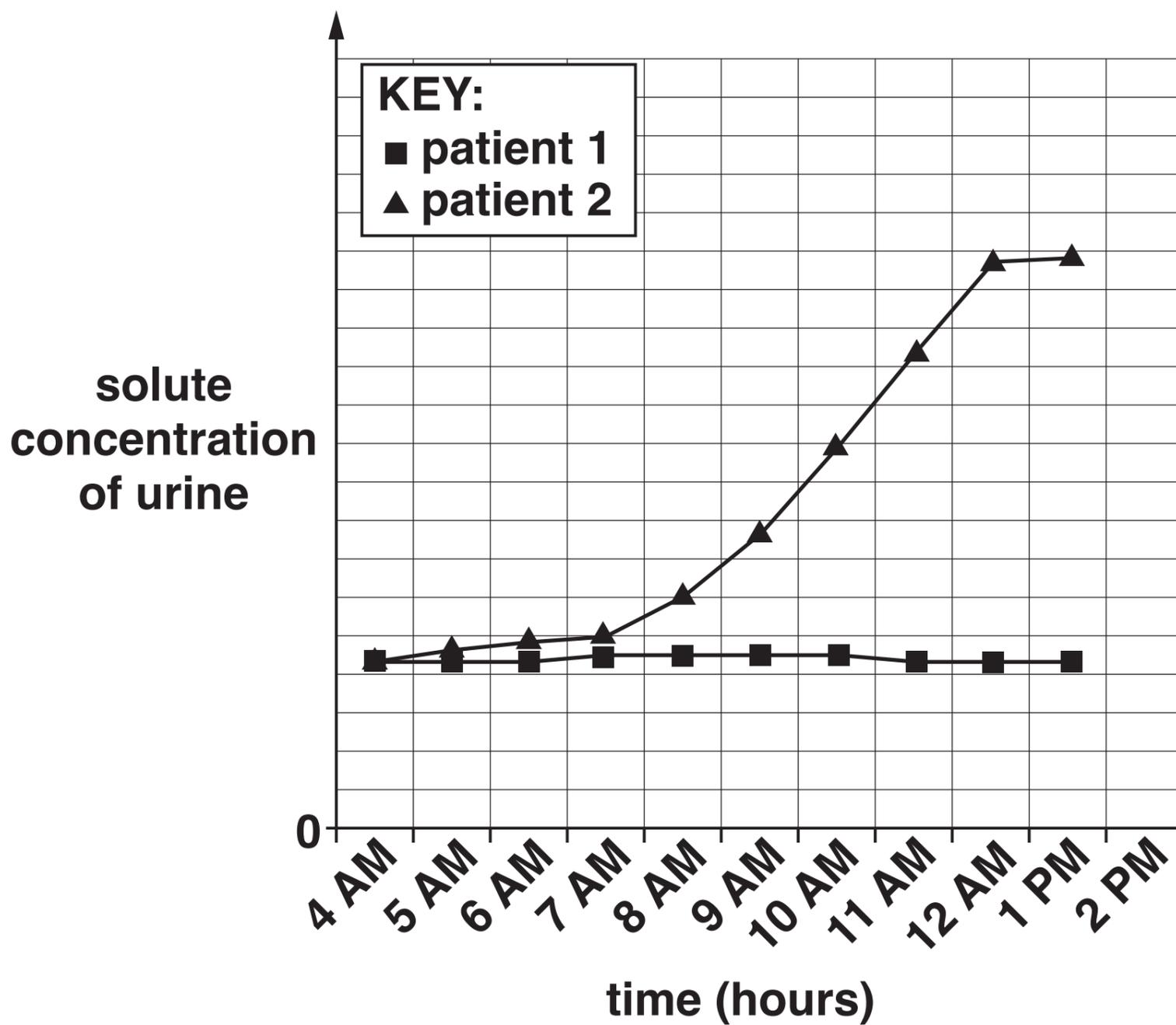
(iii) The solute concentration of each urine sample is measured.

Suggest ONE further measurement that should be carried out on the urine samples.

_____ [1]

(iv) Fig. 1.3 shows the results of a water deprivation test carried out on two different patients.

Fig. 1.3



Which of the two patients would be diagnosed with diabetes insipidus?

Use the information in Fig. 1.3 and your biological knowledge to explain your answer.

[3]

[TOTAL: 15]

2 Muscle fibres must be stimulated by a motor neurone before they contract.

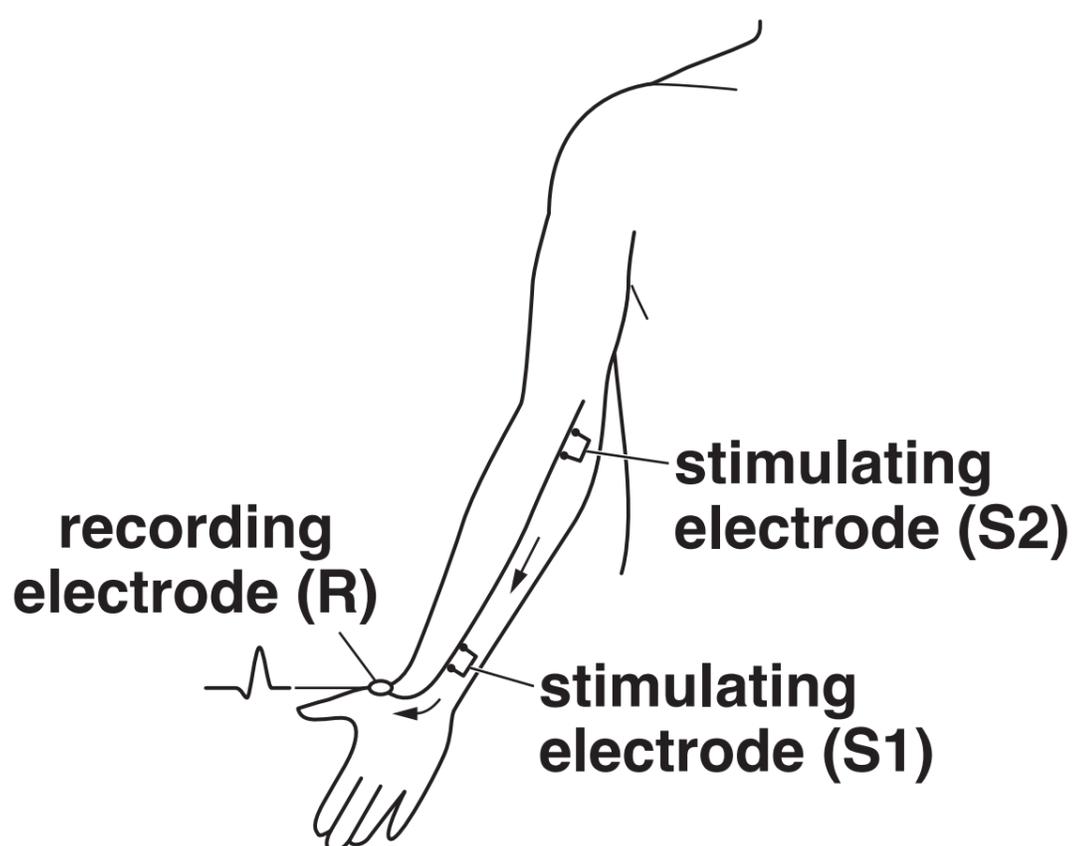
(a) What TYPE of motor neurone stimulates contraction in a skeletal muscle?

_____ [1]

(b) A nerve conduction velocity (NCV) test can be carried out to measure the speed of conduction of a nerve impulse.

An NCV test on a nerve in the arm is shown in Fig. 2.1.

Fig. 2.1



Two stimulating electrodes (S1 and S2) are attached to the skin above the nerve.

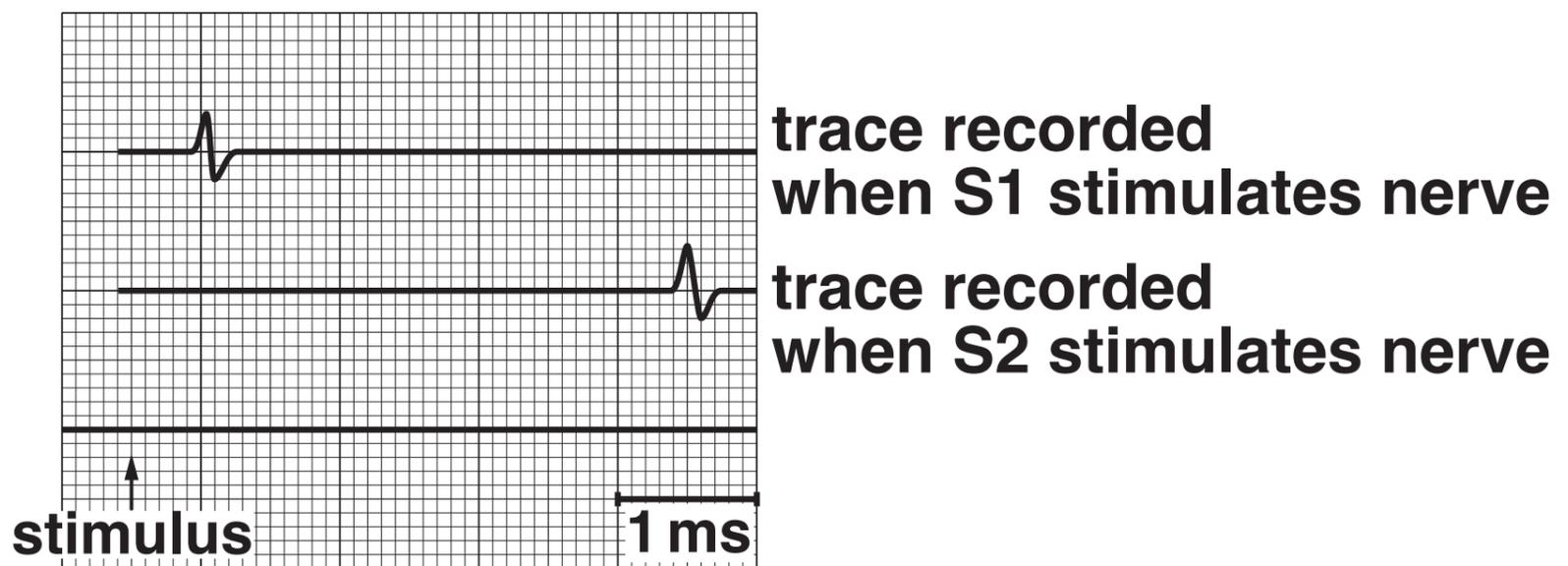
The distance (D) between the stimulating electrodes is measured.

The first electrode (S1) stimulates the nerve and the resulting electrical response is recorded by the recording electrode (R).

The second electrode (S2) stimulates the nerve and the electrical response is also recorded by R.

An NCV trace is obtained, such as that shown in Fig. 2.2.

Fig. 2.2



- (i) The nerve conduction velocity (NCV) is calculated using the formula:

$$\text{NCV} = \frac{D}{T_{S2} - T_{S1}}$$

Where:

D is the distance between the stimulating electrodes
 T_{S1} is the time taken between the stimulus given at S1 and the wave recorded at R

T_{S2} is the time taken between the stimulus given at S2 and the wave recorded at R.

Calculate the nerve conduction velocity, assuming D is 20 cm.

Show your working. Give your answer to the NEAREST WHOLE NUMBER.

Answer = _____ mm ms⁻¹ [2]

(iii) NCV tests carried out on newborn babies give values that are only half of those found in adults.

Suggest a reason for this observation.

[1]

- (c) The conduction velocity in neurones is thought to decrease as a person ages.**

In a study of the effect of ageing on SENSORY neurones, NCV tests were carried out on 1000 people. The test was repeated five years later on 500 people from the original group. Two different nerves were tested on both arms for each subject.

The following controls were in place during the study:

the temperature of the skin surface was kept at 32 °C

subjects diagnosed with diabetes mellitus were excluded from the study.

- (i) Explain why it was important to keep skin temperature constant AND suggest why a temperature of 37 °C was NOT selected.**

[2]

- (ii) Suggest a reason why subjects with diabetes mellitus were excluded from this study.**

[1]

(iii) The results from the NCV study are given in Table 2.1.

Table 2.1

NERVE TESTED	MEAN VELOCITY CHANGE AFTER 5 YEARS (ms^{-1})	PROBABILITY (P-value)	CONFIDENCE LIMITS (ms^{-1})
Nerve 1	-1.1	$p < 0.0001$	-1.49, -0.62
Nerve 2	-0.7	$p = 0.002$	-1.15, -0.27

What TWO conclusions about conduction velocity in neurones can be drawn from ALL the information shown in Table 2.1?

[2]

[TOTAL: 16]

3 A successful organ transplant usually depends on a good genetic match between the organ donor and the recipient to avoid the transplanted organ being rejected.

(a) Complete the following paragraph about the major histocompatibility (MHC) system by inserting the most appropriate term.

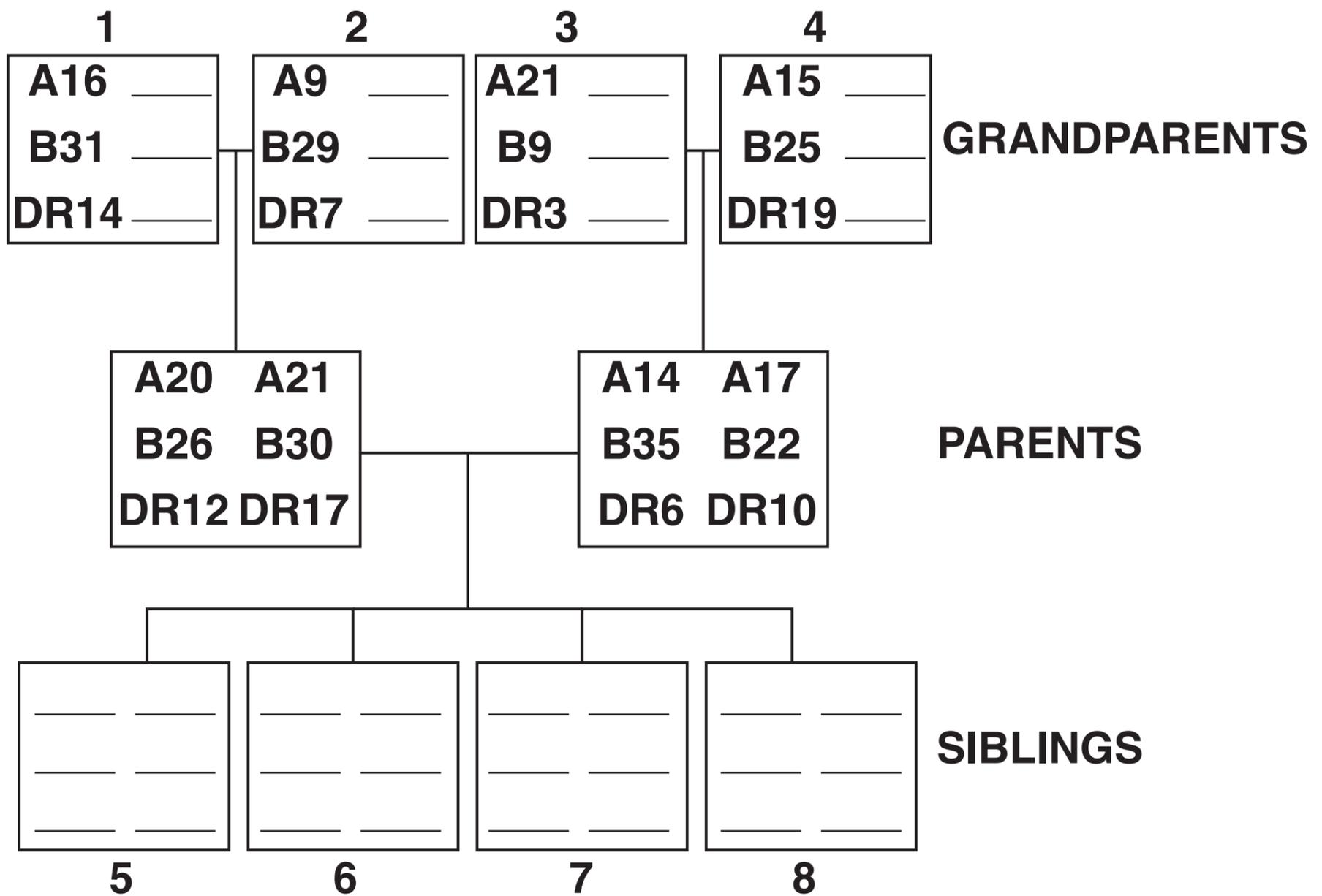
There are six major genes controlling the MHC complex located on chromosome 6. Each gene codes for a _____, which is expressed on the cell surface membrane and acts as an _____. Each of the six genes has many different alleles.

The gene _____ are close together on chromosome 6, making it unlikely that _____ will occur between them during prophase 1 of _____. This means that one complete set of alleles or HAPLOTYPE will be inherited from each parent. The MHC alleles are said to be _____, as the alleles from both haplotypes are expressed in the phenotype. [6]

(b) For transplants of organs such as kidneys, the important genes are MHC genes A, B and DR. A has approximately 25 different alleles, B has 40 and DR has 20.

Fig. 3.1 shows a pedigree diagram for the inheritance of haplotypes in three generations of one family.

Fig. 3.1



(i) Complete Fig. 3.1 to show the haplotypes of the grandparents AND the four possible haplotypes of the siblings.

The answer to this question should be written on Fig. 3.1.

[3]

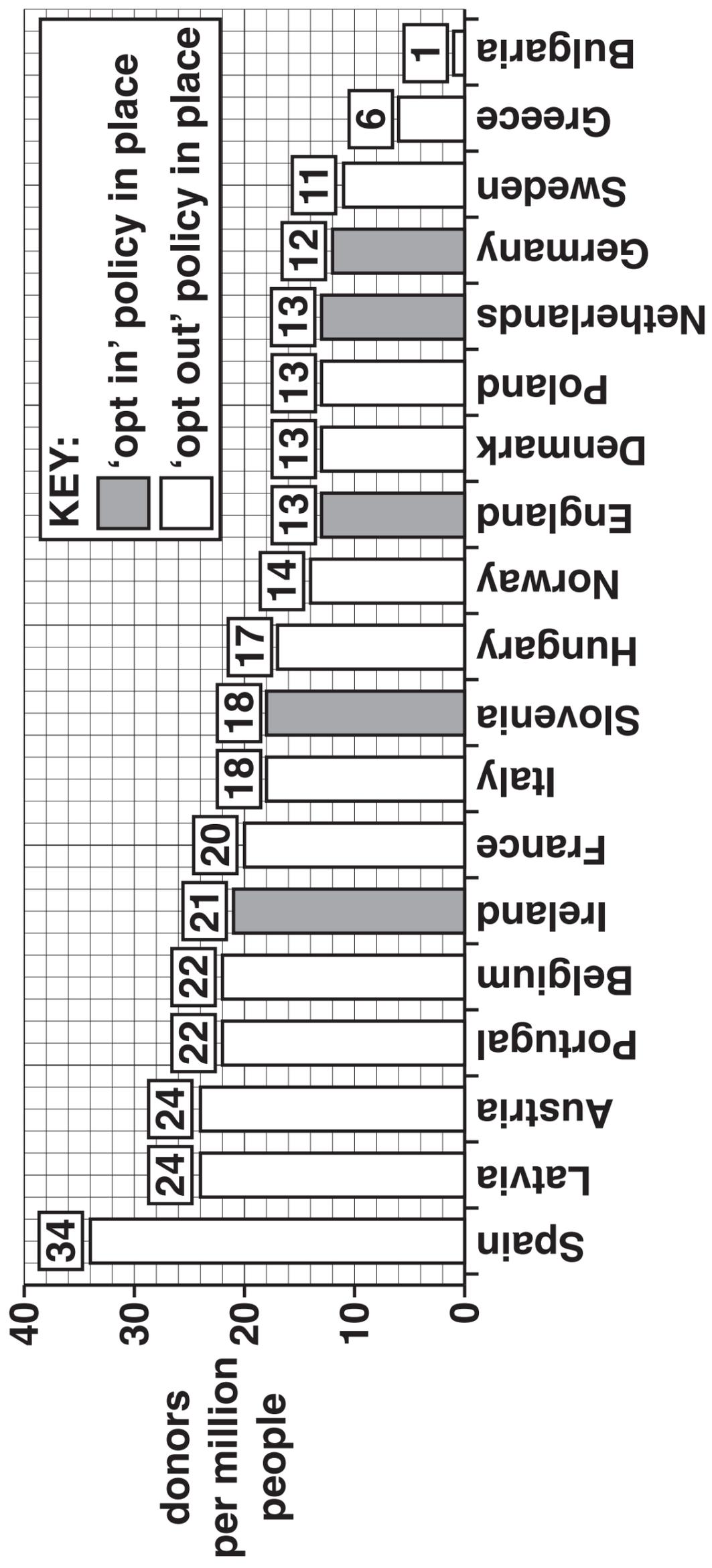
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(d) The current policy for organ donation in England is an 'opt in' approach. People choose to become registered as an organ donor and specify that they consent to their organs being used in transplant surgery.

Some European countries have an 'opt out' policy where people are PRESUMED to be willing to donate their organs unless they specify that they do NOT wish to.

Fig. 3.2 opposite is a bar chart showing the number of people donating organs in different countries, some of which have 'opt in' policies, such as England, and some of which have 'opt out' policies.

Fig. 3.2



(ii) State ONE advantage of using a kidney transplant as an alternative to haemodialysis for treating kidney failure.

[1]

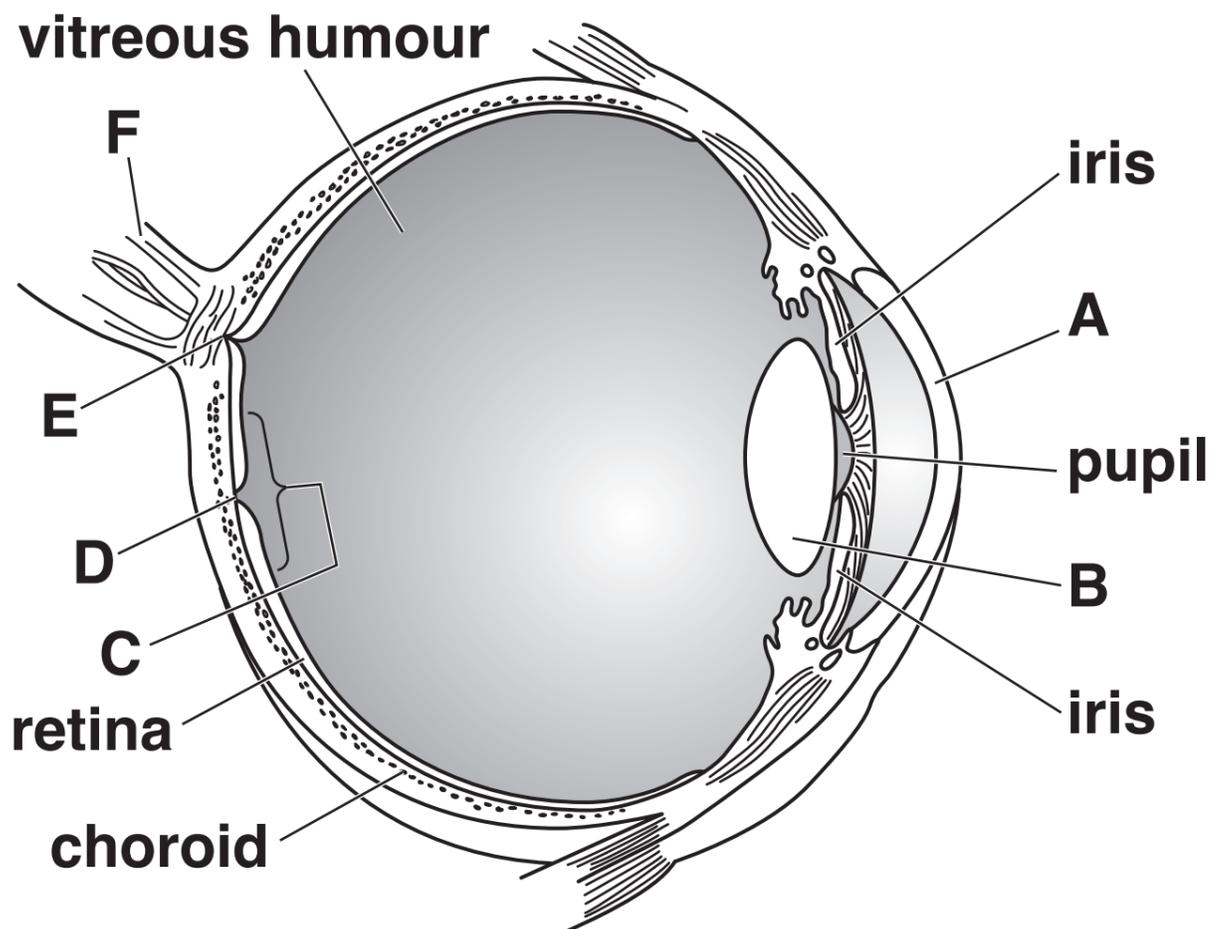
[TOTAL: 20]

[TOTAL: 8]

5 (a) Partial sight or blindness can occur as a result of damage to several different parts of the eye.

Fig. 5.1 shows a diagram of the eye.

Fig. 5.1



Complete the table by inserting the appropriate letter or letters from Fig. 5.1.

DESCRIPTION	LETTER(S)
The region that is damaged if glaucoma is not treated	
The region where a cataract can form	
The region of the retina where only cones are found	
The region of the retina where both rods and cones are found	
The region of the retina where no rods or cones are found	

[5]

(b) Choroideremia is an inherited sex-linked recessive condition that results in degeneration of the choroid layer in the eye. This condition leads to a gradual breakdown of the retina and eventual blindness.

Using the normal conventions for constructing genetic diagrams and the letters E and/or e, choose appropriate symbols to represent:

(i) the allele for choroideremia

_____ [1]

(ii) the possible genotype(s) of a person who develops choroideremia

_____ [1]

(iii) the genotype of a carrier of choroideremia.

_____ [1]

(c) The gene involved in choroideremia codes for a protein called REP-1.

Many of the gene mutations that cause choroideremia result in the formation of a protein that is much smaller than normal REP-1. This smaller protein is known as a TRUNCATED PROTEIN.

(i) State the ORGANELLE in the cell where the following occurs:

a complementary RNA copy of the gene is

synthesised _____

the REP-1 protein is synthesised

[2]

(ii) Suggest HOW a mutation in the REP-1 gene could lead to the formation of a truncated protein.

[2]

(d) A phase 1 clinical trial was carried out using gene therapy to treat choroideremia. An outline of the method used is given below.

A complementary DNA (cDNA) copy of the REP-1 gene was inserted into a viral vector.

The retina of the patient was detached to expose the choroid layer.

A fine needle was used to inject the virus into the choroid layer.

(i) Name the enzyme used to create a cDNA copy of the REP-1 gene.

[1]

(ii) What type of gene therapy has been used in the trial?

[1]

(iii) Discuss the reasons why genetic diseases such as choroideremia are good choices for treatment using gene therapy.

[3]

[TOTAL: 17]

6 The World Health Organisation has identified ways that alcohol can cause harm. One of these ways relates to the **QUALITY** of the alcohol consumed.

The term 'alcohol' covers a range of chemicals. The alcohol present in beers, wines and spirits is ethanol. Ethanol is produced by yeast cells from the breakdown of carbohydrates.

(a) Name the metabolic pathway that produces ethanol.

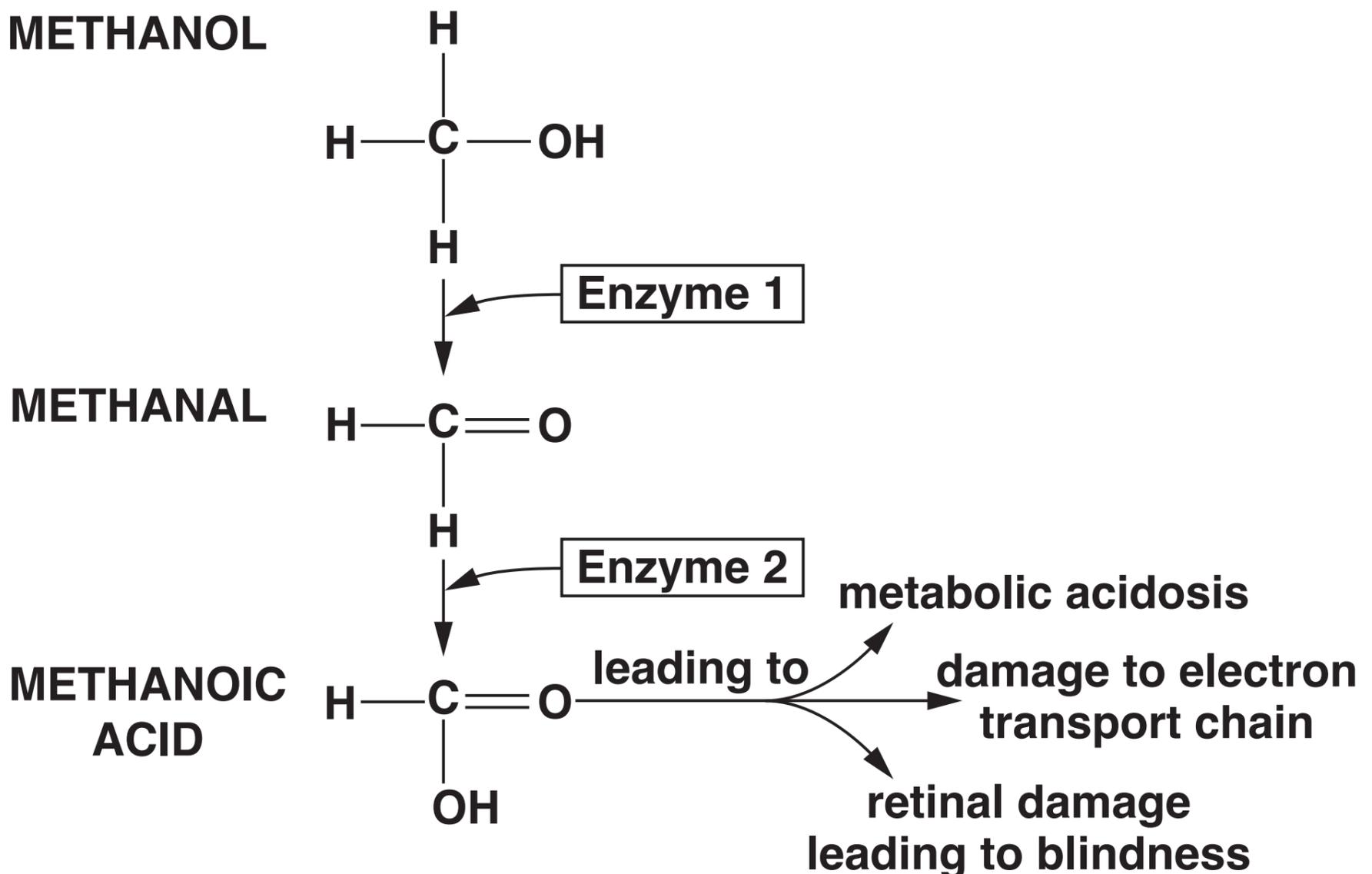
_____ [1]

(b) Methanol is an alcohol that can contaminate homemade or illegally produced alcoholic drinks.

Methanol is not toxic, but it is metabolised in liver cells to produce toxic substances such as **METHANAL** and **METHANOIC ACID**.

The metabolic pathway for methanol breakdown is shown in Fig. 6.1.

Fig. 6.1



- (i) Metabolic acidosis refers to the build up of organic acids in the blood plasma.**

The methanoic acid formed from the metabolism of methanol is only slowly broken down, leading to a build up of this acid in the blood plasma.

Using the information in Fig. 6.1, suggest one further organic acid that also builds up in the blood plasma. Explain your suggestion.

Name of acid _____

Explanation _____

_____ [3]

- (ii) Enzyme 1 in Fig. 6.1 is alcohol dehydrogenase. Alcohol dehydrogenase also breaks down ethanol in liver cells.**

Using your knowledge of enzyme activity, suggest why administering ethanol is one method of treating methanol poisoning.

_____ [2]

- (iii) There are several different alleles that code for the production of alcohol dehydrogenase in humans. The activity of the enzyme varies with the allele. Some of these alleles are linked to an increased risk of alcohol dependency.

What type of dependency is associated with variation in the alleles for alcohol dehydrogenase?

Explain your answer.

[1]

[TOTAL: 7]

7 Different healthcare professionals are involved in the diagnosis and treatment of medical conditions and diseases.

(a) Complete Table 7.1 by inserting appropriate examples of the following in each row:

the named medical condition or disease

the healthcare professional

a diagnostic test

a treatment.

Table 7.1

MEDICAL CONDITION OR DISEASE	HEALTHCARE PROFESSIONAL	DIAGNOSTIC TEST	TREATMENT
	radiographer	DEXA scan	calcium supplements
anaemia	nurse specialising in chronic kidney disease		
		pedigree analysis	
	paramedic	take core body temperature	application of ice packs

[6]

(b) The Diabetes Specialist Nurse (DSN) is just one healthcare professional who will be involved in the treatment of patients with type 1 and type 2 diabetes.

A DSN must be able to provide and give advice on INJECTABLE therapies for HYPERGLYCAEMIA. One of the injectable therapies used to treat TYPE 2 diabetes uses GLP-1.

GLP-1 is a small peptide produced naturally by the gut.

GLP-1 has several effects including:

decreasing glucose release from the liver

decreasing gastric (stomach) emptying

increasing insulin release from the pancreas

decreasing glucagon release from the pancreas.

(i) Suggest why GLP-1 is injected rather than taken orally.

_____ [1]

(ii) In what form is glucose normally stored in the liver?

_____ [1]

(iii) Suggest how a delay in the emptying of the stomach could improve the management of type 2 diabetes.

_____ [1]

(iv) Name the cells in the pancreas where GLP-1 acts.

_____ [2]

(c) A DSN must also be able to provide and give advice on ORAL therapies for hyperglycaemia and hypoglycaemia.

(i) One of the drugs used in oral therapies for HYPERGLYCAEMIA in TYPE 2 diabetes targets the enzyme that normally breaks down GLP-1.

Suggest how the action of this drug on the enzyme lowers hyperglycaemia.

_____ [1]

(ii) Explain why this drug is NOT used in the treatment of hyperglycaemia in TYPE 1 diabetes.

_____ [1]

(iii) State how HYPOGLYCAEMIA would be treated orally in type 1 diabetes.

_____ [1]

- (d) In addition to monitoring injectable and oral therapies, and interpreting blood and urine test results, a Diabetes Specialist Nurse (DSN) will also be involved in other aspects of the long-term care of people with diabetes.

Discuss the role of the DSN in the long-term care of people with diabetes OTHER THAN in monitoring different therapies and interpreting blood and urine test results.

[3]

[TOTAL: 17]

END OF QUESTION PAPER



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