RECOGNISING ACHIEVEMENT

Oxford Cambridge and RSA Examinations

Advanced Subsidiary General Certificate of Education

HUMAN BIOLOGY
Genetics, Homeostasis and Ageing

2867

Specimen Paper 2003

Additional materials: Ruler (cm/mm)
Electronic calculator

TIME 2 hours

Candidate Name	Centre Number	Candidate Number

INSTRUCTIONS TO CANDIDATES

- Write your name, Centre number and candidate number in the spaces above.
- Write your answers, in blue or black ink, in the spaces provided on the question paper.
- Answer **all** the questions.
- Read each question carefully and make sure you know what you have to do before starting your answer.

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 120.
- You may use an electronic calculator.
- You are advised to show all the steps in any calculations.
- *You will be awarded marks for the quality of written communication where an answer requires a piece of extended writing.*

FOR EXAMINER'S USE		
Question number	**Max.**	**Mark**
1	**19**	
2	**15**	
3	**18**	
4	**18**	
5	**16**	
6	**21**	
7	**13**	
TOTAL	**120**	

1 The number of diagnosed cases of Type 2 diabetes is increasing faster than any other chronic disease in the United Kingdom.

(a) **(i)** State **two** factors which increase the risk of developing Type 2 diabetes.

1 _____

2 _____ **[2]**

(ii) Describe **four** differences between Type 1 and Type 2 diabetes.

_____

_____

_____

_____

_____ **[4]**

(b) **Fig. 1.1** shows the effect of **three** successive oral doses of glucose on the blood glucose concentration of two individuals, **A** and **B**.

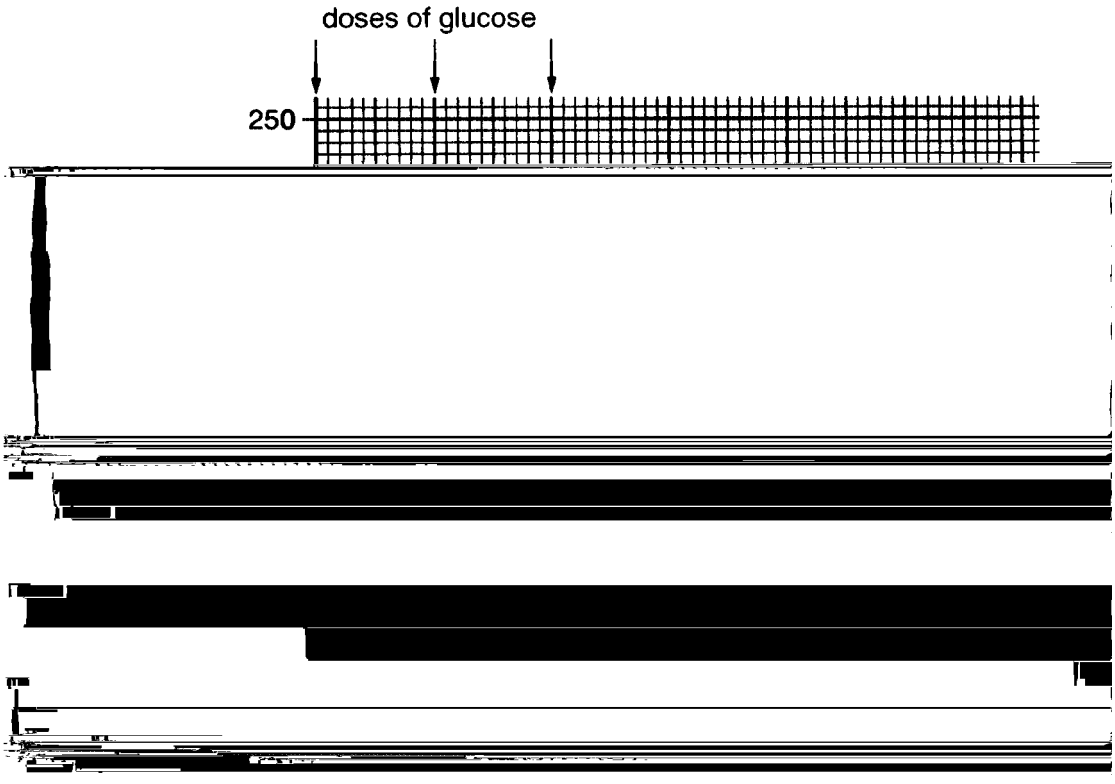

Fig. 1.1

1 **(b)** Use the information given on **Fig. 1.1** to explain the results for:

(i) Individual **A**;

[4]

(ii) Individual **B**.

[2]

(c) **Fig 1.2** shows the chemical structure of insulin. The amino acids thought to bind to the insulin receptors on cell surface membranes are shown in black.

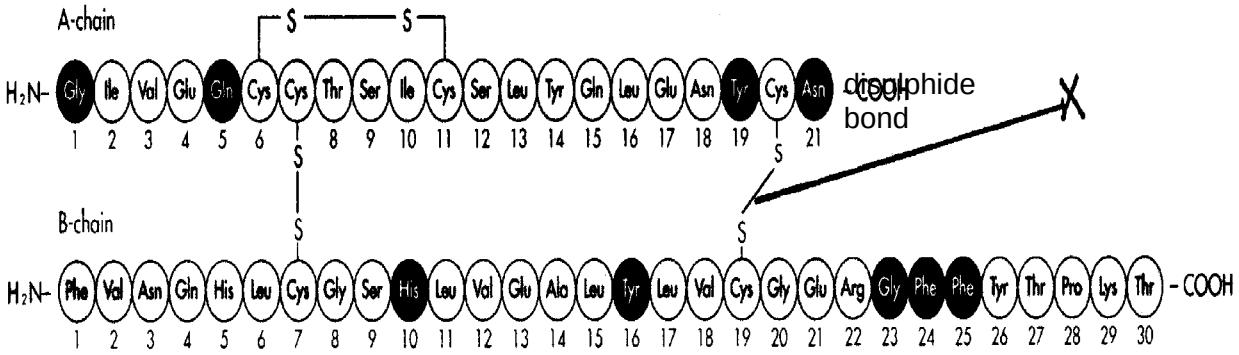

Fig. 1.2

1 **(c)** **(i)** Explain the importance of the disulphide bond labelled on **Fig. 1.2** to the function of the insulin molecule.

[3]

(ii) Explain what is meant by the phrase *'insulin receptors on cell surface membranes'*, and describe how these receptors are involved in the action of insulin in lowering blood glucose concentration.

[4]

2 As life expectancy increases it is likely that the number of patients with age related illnesses such as osteoporosis and osteoarthritis will increase.

(a) Explain the meaning of the following terms:

osteoporosis

[2]

osteoarthritis

[2]

2 **(b)** The measurement of bone density is one of the techniques used to monitor changes in the structure of bone due to ageing.

Fig. 2.1 shows the changes in bone density in men and women from the age of 20 years to 90 years.

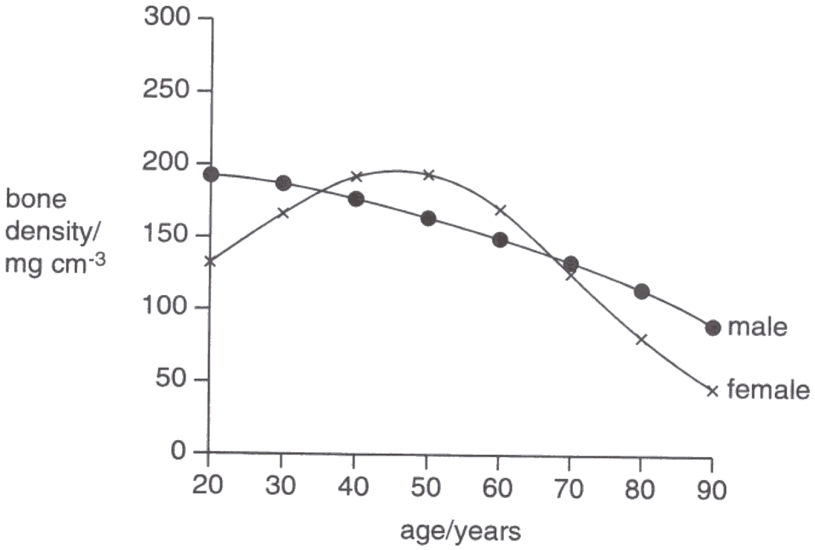

Fig. 2.1

Using the information on **Fig. 2.1**:

(i) describe how bone density changes with age in women;

_____

_____

_____

_____ **[2]**

(ii) explain **why** the curve for men differs from that for women.

_____

_____

_____

_____ **[2]**

2 **(c)** Discuss the **social** consequences for the individual of osteoporosis and describe ways in which osteoporosis can be prevented.

[7]

3 The health of the gaseous exchange system has a major effect on the performance of the human body**. Fig. 3.1** shows a diagram, drawn from a photomicrograph, of gaseous exchange tissue in the lungs.

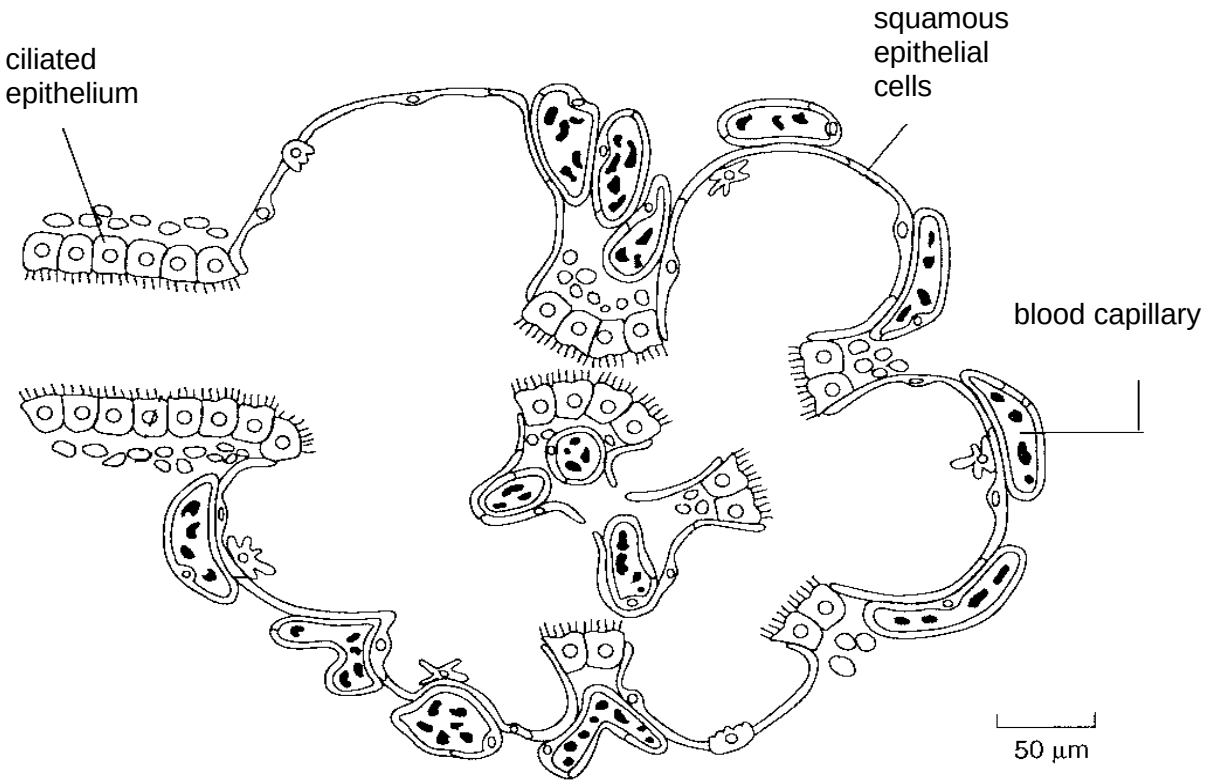

Fig. 3.1

(a) (i) Describe how the gaseous exchange tissue is adapted to its function.

[4]

3 **(a)** **(ii)** Describe the effect of ageing on the gaseous exchange system.

[5]

(b) **Fig. 3.2** summarises part of the respiratory pathway in human cells.

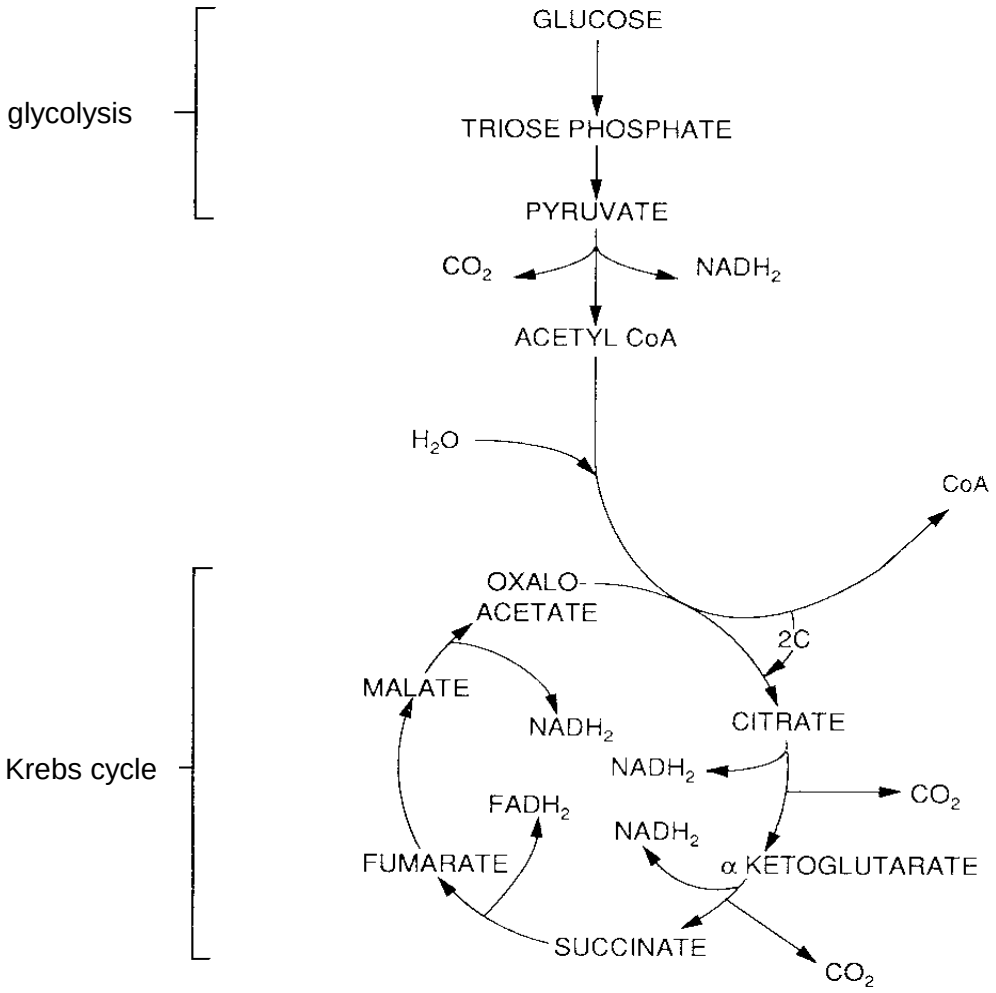

Fig. 3.2

3 (b) (i) Complete the table below by stating the precise location in the cell where glycolysis and the Krebs Cycle occur.

Stage	Precise location in cell
glycolysis	
Krebs cycle	

[2]

(ii) *In this question, one mark is available for the quality of written communication*.

Describe how the changes in the cardiovascular system due to ageing, will change the pathway shown in **Fig. 3.2** during exercise.

[6]

Quality of Written Communication [1]

4 A young pregnant woman consults her doctor because several members of her family have shown symptoms of a rare genetic disease which causes excessively high levels of low density lipid and cholesterol in the blood. The doctor sends the patient to a genetic counsellor who questions her about the occurrence of the disease in her family.

Fig. 4.1 shows the family pedigree of the patient, based on the information received by the genetic counsellor. The patient herself is shown as *generation* III, 1.

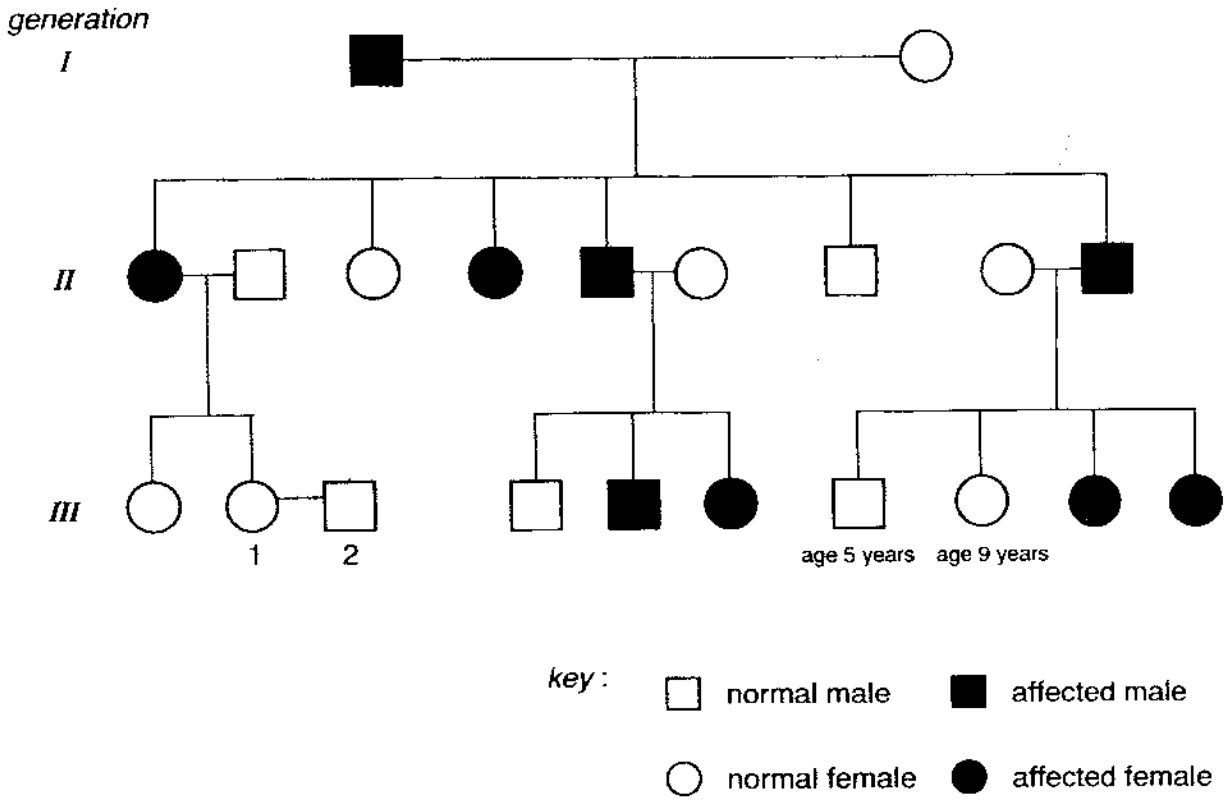

Fig. 4.1

(a) (i) State the type of inheritance shown by this condition. Give reasons for your answer.

_____

_____

_____

_____ **[2]**

4 **(a)** **(ii)** Explain, by means of a genetic diagram, the probability of the pregnant patient (generation III, 1) passing the disease on to her child.

Probability _____ **[5]**

(b) High levels of low density lipid and cholesterol in the blood are caused by a mutation of a single gene. This gene normally codes for the formation of low density lipid receptors in the endothelium of the blood vessels. These receptors normally remove low density lipid from the blood.

(i) Explain in detail how a mutation in this gene could change the structure of the receptor, preventing it removing the low density lipid from the blood.

_____

_____

_____

_____

_____

_____ **[4]**

(ii) Explain why this mutation will also result in high levels of cholesterol in the blood.

_____

_____

_____ **[2]**

4 **(b)** **(iii)** Describe the likely long-term consequences of this mutation on the cardiovascular system.

[3]

(iv) Suggest a reason why this potentially fatal allele still survives in the population.

[2]

5 As life expectancy increases, the prevalence of diseases, such as Alzheimer's disease, is expected to increase. People affected by Alzheimer's disease will require specialised care.

(a) **(i)** Outline **two** consequences of Alzheimer's disease for the family of the patient.

1

2

[4]

(ii) Suggest a way in which the welfare services could help the family of the patient.

[2]

5 **(b)** The diagrams in **Fig. 5.1** show the length and appearance of the dendrites of neurones in a part of the forebrain known as the hippocampus. The neurones are from healthy adults aged 50, 60 and 70 years and from a 70 year old with Alzheimer's disease.

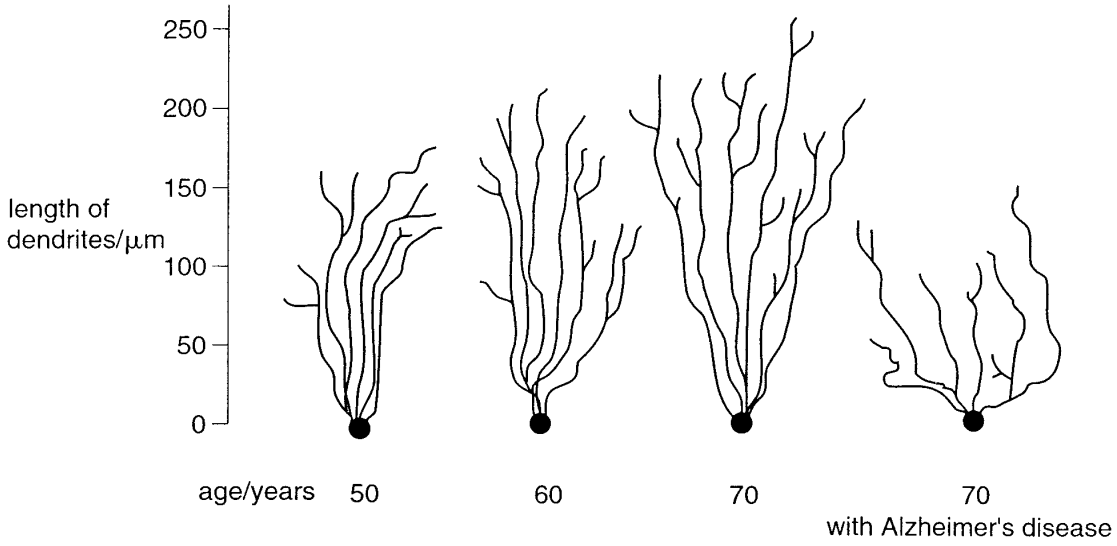

Fig. 5.1

Using the information on **Fig. 5.1**:

(i) Describe the changes in the length and appearance of the dendrites of healthy adults, with increasing age;

_____

_____

_____ **[2]**

(ii) Describe how the dendrites differ in the 70 year old with Alzheimer's disease.

_____

_____

_____ **[2]**

5 continued

Studies have shown that about 5% of neurones in the hippocampus disappear with each decade after the age of 50.

(c) For every 100 neurones present in the hippocampus at age 50, calculate, to the nearest whole number, how many will be present from age 70. Show your working.

Answer _____ **[2]**

(d) Suggest how the changes in the neurones of healthy adults, as shown in **Fig. 5.1**, are related to the loss of neurones with age.

_____

_____

_____ **[2]**

(e) Loss of neurones may also occur in the cerebellum. Suggest the likely effects of this on the body.

_____

_____

_____ **[2]**

6 Chronic kidney disease has a profound effect on the life of a patient. Diet and blood pressure have to be monitored carefully if the patient is to remain well.

(a) Caffeine, an active ingredient in coffee, causes vasodilatation of the afferent arterioles in the nephrons.

Suggest the effect that caffeine will have in the kidney on:

(i) blood flow through the glomerulus;

_____

_____ **[1]**

(ii) the rate of urine production.

_____

_____ **[1]**

6 **(b)** If the blood pressure remains too high for a long period, an individual may develop symptoms of kidney disease.

(i) State **two** symptoms of kidney disease.

1 _____

2 _____ **[2]**

(ii) Explain how prolonged high blood pressure may cause chronic kidney disease.

_____

_____

_____ **[2]**

(c) If kidney disease cannot be treated, the kidneys may fail and a kidney transplant may be required. If the transplant is to succeed, it is important that the blood group and tissue type of the donor and recipient match as closely as possible.

Explain the likely response of the immune system of the recipient if the match of the two tissues is not good.

_____

_____

_____

_____

_____ **[4]**

(d) The tissue type is determined by proteins on the cell surface membrane. These are coded for by several gene loci situated very close together on chromosome 6. Each locus has a number of different alleles. This group of loci are inherited together and is called a haplotype.

(i) State the genetic term which is used when alleles on the same chromosome are inherited together.

_____ **[1]**

6 (d) (ii) The parents of a child suffering from kidney failure have volunteered to donate a kidney to their child. Their haplotypes and those of their child are shown in **Fig. 6.1**.

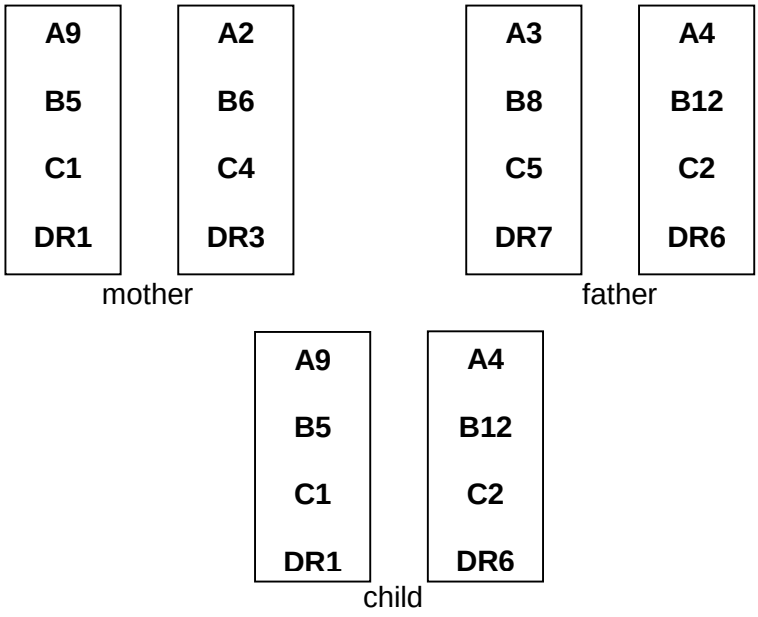

Fig. 6.1

Explain, by means of a genetic diagram, why the child has a better chance of a successful transplant from the other children in the family than from either parent.

[3]

6 **(e)** *In this question, one mark is available for the quality of written communication*.

There are approximately two thousand kidney transplant operations carried out in the United Kingdom each year, but there are twice as many patients waiting for a suitable donated kidney.

Discuss the social and moral issues associated with the use of kidney transplants.

[6]

Quality of Written Communication [1]

7 Genetic engineering using recombinant DNA technology is a promising area of medical research into treating, and possibly curing, genetic disease. Many bacteria synthesise restriction enzymes, which cut viral DNA at specific nucleotide base sequences. This protects the bacteria from viral attack. The sequence of nucleotide bases where one restriction enzyme cuts is shown in **Fig. 7.1**.

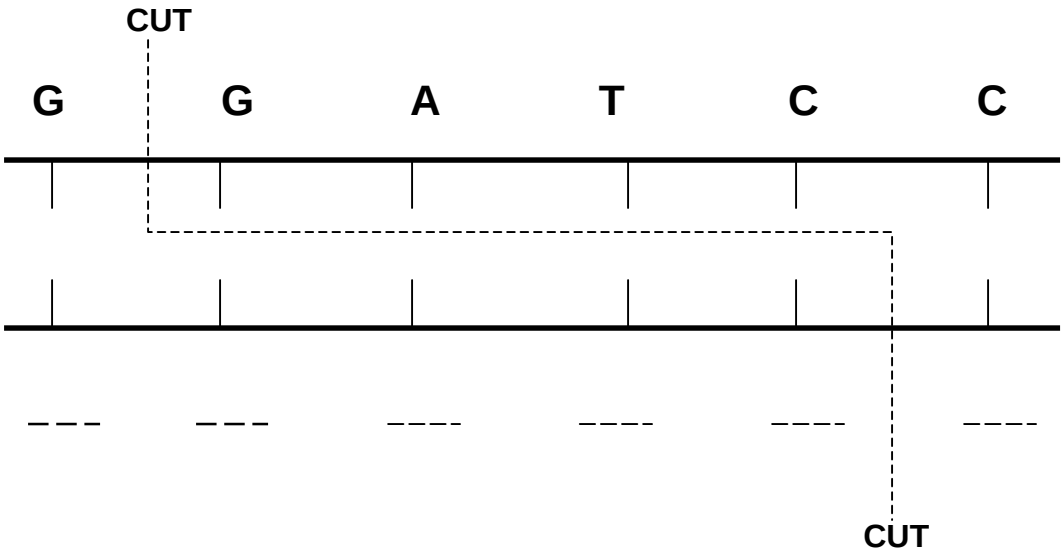

Fig. 7.1

(a) Complete **Fig. 7.1** by writing in the letters for the nucleotide bases belonging to the complementary chain. **[1]**

(b) Explain why the sequence of bases in DNA are in the form of a 'triplet code'.

_____

_____

_____

_____ **[2]**

(c) **(i)** Explain how restriction enzymes recognise specific base sequences.

_____

_____

_____

_____ **[2]**

7 **(c)** **(ii)** Explain why it is necessary to maintain a constant temperature in experiments involving restriction enzymes.

[2]

(d) Explain what is meant by recombinant DNA and describe how restriction enzymes are used to produce recombinant DNA.

[6]

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MARK SCHEME

ADVICE TO EXAMINERS ON THE ANNOTATION OF SCRIPTS

1 Please ensure that you use the **final** version of the Mark Scheme.
You are advised to destroy all draft versions.

2 Please mark all post-standardisation scripts in red ink. A tick (✓) should be used for each answer judged worthy of a mark. Ticks should be placed as close as possible to the point in the answer where the mark has been awarded. The number of ticks should be the same as the number of marks awarded. If **two** (or more) responses are required for one mark, use only **one** tick. Half marks (½) should never be used.

3 The following annotations may be used when marking. **No comments should be written on scripts unless they relate directly to the mark scheme. Remember that scripts may be returned to Centres.**

X	=	incorrect response (errors may also be underlined)
^	=	omission mark
bod	=	benefit of the doubt (where professional judgement has been used)
ecf	=	error carried forward (in consequential marking)
con	=	contradiction (in cases where candidates contradict themselves in the same response)
sf	=	error in the number of significant figures

4 The marks awarded for each part question should be indicated in the margin provided on the right hand side of the page. The mark total for each question should be ringed at the end of the question, on the right hand side. These totals should be added up to give the final total on the front of the paper.

5 In cases where candidates are required to give a specific number of answers, (e.g. 'give three reasons'), examiners should look at the responses given and make a judgement as to whether they are correct, incorrect or 'neutral'. They should then mark the first answer(s) given (unless they are judged to be 'neutral') up to the total number required. Strike through the remainder. In specific cases where this rule cannot be applied, the exact procedure to be used is given in the mark scheme.

6 Correct answers to calculations should gain full credit even if no working is shown, unless otherwise indicated in the mark scheme. (An instruction on the paper to 'Show your working' is to help candidates, who may then gain partial credit even if their final answer is not correct.)

7 Strike through all blank spaces and/or pages in order to give a clear indication that the whole of the script has been considered.

8 An element of professional judgement is required in the marking of any written paper, and candidates may not use the exact words that appear in the mark scheme. If the science is correct **and** answers the question, then the mark(s) should normally be credited. If you are in doubt about the validity of any answer, contact your Team Leader/Principal Examiner for guidance.

Abbreviations, annotations and conventions used in the Mark Scheme	/	=	alternative and acceptable answers for the same marking point
	;	=	separates marking points
	NOT	=	answers which are not worthy of credit
	( )	=	words which are not essential to gain credit
	____	=	(underlining) key words which **must** be used to gain credit
	ecf	=	error carried forward
	AW	=	alternative wording
	ora	=	or reverse argument

Question	Answer	Mark
1(a)(i)	genetic potential; obesity; high refined carbohydrate diet / AW *diet must be qualified* *Accept the first answer given on each answer line (unless the first is judged to be 'neutral'). If all the answer(s) given on one answer line are neutral, then look for a second correct answer on another line.*	**2 max**
1(a)(ii)	*Type 1* / *Type 2* less common / prevalent / 10-25% cases; / more common / prevalent / 75 - 90% cases; slowest rate increase / incidence; / fastest rate increase/incidence; sudden onset; / gradual onset; early onset; / usually late onset; insulin dependent; / diet dependent; no insulin produced; / some insulin produced; not inherited directly; / strong family trend / AW; inflammation Islets; / no inflammation Islets; beta cells destroyed; / beta cells retain some function; antibodies to Islets present; / no antibodies to Islets present. *accept first **four** answers only unless agreed as neutral.*	**4 max**
1(b)(i)	glucose absorbed into blood (causing increase in blood glucose); ref to insulin secretion; from beta cells in Islets of Langerhans; delay as insulin concentration rises / AW; glucose converted to glycogen (so glucose concentration falls); stored as fat; increase respiration / ATP production; *R energy;* increased permeability of cell membranes / liver cells to glucose; ref to insulin already present when subsequent doses given; after 3 hours, (blood) concentration returns to normal; accurate figures to illustrate any point using both axes; ® all refs to glucagon.	**4 max**
1(b)(ii)	blood glucose continues to rise / remains high; ® *increases unqualified* no / too little/not enough, insulin produced; B is diabetic/diabetes mellitus; some respired/excreted in urine; figures must include glucose concentration and time, units correct; AVP; eg ref to glucagon/decreased sensitivity to insulin.	**2 max**

Question	Answer	Mark
1(c)(i)	joins polypeptide chains; holds / forms the tertiary structure / AW; specific / precise, 3D / globular structure; matches / complements the structure;	3 max
1(c)(ii)	protein receptor; complements shape of insulin molecule / when insulin locks on; stimulates vesicles, with glucose carrier proteins / AW; to move to cell membrane; and fuse with it; increasing permeability to glucose; glucose moves out of blood and into cell / AW;	4 max
	Total mark:	**19**
2(a)	*osteoporosis* decrease in bone density; decrease in calcium salts / mobilisation calcium; brittle bones / fractures; related to decrease in oestrogen / lack of exercise;	2 max
	osteoarthritis destruction of cartilage on bone ends; loss of flexibility in ligaments; erosion / friction of bone end; decrease in synovial fluid;	2 max
2(b)(i)	women have less mass than men at 20 years; increases until 40 - 50 years; when starts to decline rapidly; at 70y decreases below that of men / less than at 20 years; figs from both axes in support.	2 max
2(b)(ii)	bones have greater mass at 20 years, as bigger; women protected until the menopause / AW; mass shows steady decrease with age because not protected by oestrogen at any stage; to prevent mobilisation calcium / AW; figs from both axes in support.	2 max
2(c)(i)	loss of independence; pain from fractures; decreased mobility; fear of falling / loss of confidence; increased need for walking aids; increased intake of calcium to increase bone mass; increased intake vitamin D to increase calcium absorption; HRT / oestrogen supplements to inhibit parathormone / mobilisation Ca^{2+}; bisphosphonate to inhibit osteoclasts / bone resorption cells; calcitonin supplement to inhibit osteoclasts / bone resorption cells; AVP;;	7 max
	Total mark:	**15**

Question	Answer	Mark
3(a)(i)	ciliated epithelium; waft debris to exterior / AW; squamous epithelium; thin, for short diffusion distance / AW; elastic, to expand / AW; moist, to assist diffusion; selectively permeable / named permeability; blood capillaries, closely applied to epithelial cells / surround epithelial cells / AW; ventilation / movement of blood, maintain diffusion gradient;	**4 max**
3(a)(ii)	lungs' ability to take up oxygen, decreases with age / AW; reduced blood supply to the lungs; tissue less efficient at oxygen uptake; smaller lung capacity; maximum O_2 uptake / V max O_2 with exercise reduced; less power from intercostal muscles / ribs more rigid / AW; reduced elasticity; less cilia / cilia less efficient; less mitosis so less repair / less regeneration of cells; ref' to more exposure to environmental factors; AVP;	**5 max**
3(b)(i)	cytoplasm; matrix of mitochondrion;	**2**
3(b)(ii)	reduced (arterial) blood, flow due to atheroma / arteriosclerosis / AW; blockage coronary arteries; reduced stroke volume / cardiac output; less oxygen to muscle; switch to anaerobic respiration / AW; Krebs cycle stops; FAD / NAD remains reduced; no decarboxylation / dehydrogenation / CO_2 / H_2 given off; pyruvate accepts hydrogen; forms lactate; small amount / two molecules ATP; O_2 debt accumulates / EPOC; AVP;; e.g. exercise stops / muscle fatigue; EPOC / oxygen consumption remains elevated after exercise; heart rate remains high after exercise; risk of heart attack / myocardial infarct increases;	**6 max**
	QWC: clear well organised answer using specialist terms;	**1**
	Total mark:	**18**
4(a)(i)	autosomal; dominant; no sex bias / equal sex distribution; present in every generation / does not skip a generation;	**2 max**

Question	Answer	Mark
4(a)(ii)	key using suitable symbols; *award only for upper case for mutant allele* parent AA / Aa x aa A *ecf throughout to max 2* correct gametes; correct genotypes linked with correct phenotypes; 0.5 probability that pregnant female has mutant allele; 0.5 probability that passed on to child; (probability therefore 0.5 x 0.5) = 0.25;	**5 max**
4(b)(i)	change in base sequence, changes amino acid sequence; ref to transcription / translation / mRNA; different amino acids in primary structure; fold differently in tertiary structure; ref to bonds;	**4 max**
4(b)(ii)	different shape does not complement shape of low density lipid / AW; cholesterol is carried by low density lipid / AW; as lipoprotein;	**2 max**
4(b)(iii)	attached to fatty acid; LDLs dump cholesterol on wall of artery / AW; roughened wall becomes fibrous / AW; ref' to phagocytes / foam cells / free radicals; platelets stick; releases clotting factors; reduces blood flow; plaque / clot blocks artery; atherosclerosis; tissue dies;	**3**
4(b)(iv)	symptoms do not show until after children produced / late onset after childbirth / AW; no natural selection;	**2**
	Total mark:	**18**
5(a)(i)	dependence of patient; loss of personal freedom; loss of job / income; dependence on state; loss of memories / personality of patient; controlling adult with childlike behaviour; psychological adjustment; AVP;; *Accept the first answer given on each answer line (unless the first is judged to be 'neutral'). If all the answer(s) given on one answer line are neutral, then look for a second correct answer on another line.*	**4 max**
5(a)(ii)	respite care, qualified / AW; help with lifting; regular carers; financial help; help with special equipment, qualified;	**2 max**

Question	Answer	Mark
5(b)(i)	dendrites get longer as age increases; dendrites get more branches as age increases; ® more dendrites	**2**
5(b)(ii)	dendrites are shorter than in healthy 70 year old; dendrites shorter than all healthy age groups; fewer branches on dendrites;	**2 max**
5(c)	$100 - \frac{10 \times 100}{100}$; = 90; *OR* correct answer only;;	**2**
5(d)	as neurones decrease, existing neurones grow longer / grow more branches / AW; to make more / replace lost, connections;	**2**
5(e)	poor processing of information; poor co-ordination of muscle movement / walking; ® control poor balance; poor fine (muscle) movement / muscle tone / adjustment of postural muscles;	**2 max**
	Total mark:	**16**
6(a)(i)	it will increase it;	**1**
6(a)(ii)	it will increase it;	**1**
6(b)(i)	protein in urine; high blood urea; loss of plasma proteins from blood / AW; oedema; high angiotensin level; imbalance ions / named / AW; AVP; *Accept the first answer given on each answer line (unless the first is judged to be 'neutral'). If all the answer(s) given on one answer line are neutral, then look for a second correct answer on another line.*	**2 max**
6(b)(ii)	basement membrane selectively permeable membrane; normally retains substance greater than 68 000 RMM; high blood pressure tears membrane; causes kidney to secrete renin; activates angiotensin; a hormone which increase blood pressure further / positive feedback effect;	**2 max**

Question	Answer	Mark
6(c)	foreign protein / antigens; recognised, by APC; presented to / activates, T4 / T helper cell; secretes cytokinins; stimulate B cell division / to form plasma cells; clonal selection; Tc cells divide by mitosis; attack foreign cells; tissue dies / killed / rejection; AVP;	**4 max**
6(d)(i)	linkage;	**1**
6(d)(ii)	*A genetic diagram or as text* gametes correctly indicated; i.e. mother A9 B5 C1 DR1 X father A3 B8 C5 DR7 A2 B6 C4 DR3 A4 B12 C2 DR6 offspring; i.e. A9 B5 C1 DR1, A3 B8 C5 DR7 A9 B5 C1 DR1, A4 B12 C2 DR6 A2 B6 C4 DR3, A3 B8 C5 DR7 A2 B6 C4 DR3, A4 B12 C2 DR6 .25 / 25% / 1 in 4 chance of having same tissue type / genotype as recipient; parents can only match five out of eight alleles; risk of rejection from parents / less risk from sibling;	**3 max**
6(e)	not enough donors; risk of paying living donor; exploitation of vulnerable living donors; pressure on relative as living donor; pressure on relatives of dying donor; cost of operation / medication when resources limited; need to take medication throughout life; psychological problems for recipient;	**6 max**
	QWC: legible text, accurate spelling, punctuation and grammar;	**1**
	Total mark:	**21**
7(a)	C C T A G G;	**1**
7(b)	only a code of <u>three</u> bases; could include all 20 / 21 amino acids; A *4*[3] into polypeptide;	**2 max**
7(c)(i)	active site of restriction enzyme; complements shape specific bases; forms enzyme substrate complex / binds to DNA;	**2 max**

Question	Answer	Mark
7(c)(ii)	enzymes have an optimum temperature; at which work fastest; works slowly / inhibited, at low temperatures / AW; denatured ,above optimum temperature / if temperature too high;	2 max
7(d)	DNA incorporates foreign DNA / DNA from another source; cuts donor DNA at a specific site; to incorporate gene / allele required; symmetrical site / palindromic; leaves sticky ends; cut host DNA with same restriction enzyme; leaves complementary sticky ends; these fuse; hydrogen bond; sugar phosphate backbone sealed with DNA ligase;	6 max
	Total mark:	**13**
	PAPER TOTAL:	**120**