

**Human Biology**

Advanced GCE

Unit **F225**: Genetics, Control and Ageing

**Mark Scheme for June 2013**

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This mark scheme is published as an aid to teachers and students, to indicate the requirements of the examination. It shows the basis on which marks were awarded by examiners. It does not indicate the details of the discussions which took place at an examiners' meeting before marking commenced.









All examiners are instructed that alternative correct answers and unexpected approaches in candidates' scripts must be given marks that fairly reflect the relevant knowledge and skills demonstrated.

Mark schemes should be read in conjunction with the published question papers and the report on the examination.

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Annotation	Meaning
/	alternative and acceptable answers for the same marking point
(1)	separates marking points
not	answers which are not worthy of credit
R	answers which are not worthy of credit
ignore	statements which are irrelevant
allow	answers that can be accepted
( )	words which are not essential to gain credit
—	underlined words must be present in answer to score a mark
ecf	error carried forward
AW	alternative wording
ora	or reverse argument

SCORIS Annotation	Meaning
	correct response
	incorrect response
	benefit of the doubt
	benefit of the doubt <b>not</b> given
	error carried forward
	information omitted
	ignore
	reject



Question			Answer	Marks	Guidance			
					<i>Type of disease</i>	<i>HRT</i>	<i>PLACEBO</i>	<i>difference</i>
					COLORECTAL CANCER	10	15	5
					HIP FRACTURE	10	15	5
					CHD	37	30	7
					STROKE	30	20	10
					BREAST CANCER	39	32	7

Question		Answer	Marks	Guidance
	(d)	<p><i>hysterectomy</i> (hysterectomy is) removal of, uterus / womb ;</p> <p>would not be able to investigate endometrial cancer ;</p> <p><i>blood pressure</i> hypertension / high blood pressure, (also) increases risk of, CHD / strokes ;</p> <p><i>for either</i> this would be a confounding variable / AW <b>OR</b> <i>idea that</i> this needs to be controlled for a <u>valid</u> investigation / comparison ;</p>	3	<p><b>DO NOT CREDIT</b> if given as part of a list with e.g. ovaries <b>ACCEPT</b> 'will not get endometrial cancer'</p> <p><b>DO NOT CREDIT</b> idea of a lower risk of this cancer</p> <p><b>ACCEPT</b> a description e.g.' it would be hard to tell if it was HRT or high blood pressure or both leading to increased risk'</p> <p><b>DO NOT ACCEPT</b> 'valid' if given as part of a list with 'reliable', 'precise' or 'accurate'.</p>
		<b>Total</b>	<b>12</b>	

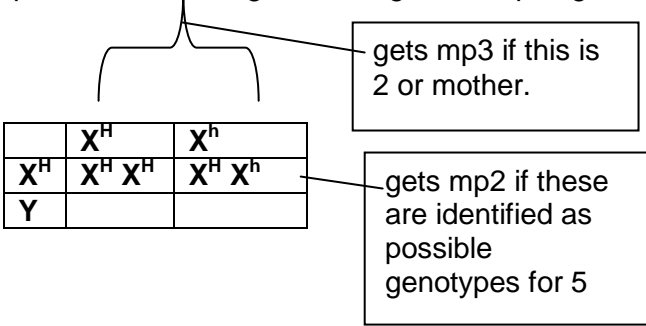
Question			Answer	Marks	Guidance												
2	(a)	(i)	phospholipid (bilayer) ;	1													
		(ii)	no, myelin sheath / Schwann cell (around axon membrane) ;  (presence of proteins for) movement of ions in <b>and</b> out (of axon) ;  presence of, sodium - potassium pumps ;	1	<b>CREDIT</b> Na <sup>+</sup> and K <sup>+</sup> for 'ions' throughout  <b>CREDIT</b> reference to <u>exchange</u> of ions  <b>IGNORE</b> reference to numbers of ions moved or direction												
	(b)		<table border="1" style="margin-left: auto; margin-right: auto;"> <thead> <tr> <th><i>depolarisation</i></th> <th><i>repolarisation</i></th> </tr> </thead> <tbody> <tr> <td>closed</td> <td>open</td> </tr> <tr> <td>open</td> <td></td> </tr> <tr> <td></td> <td>no diffusion</td> </tr> <tr> <td>some diffusion out</td> <td>rapid diffusion out</td> </tr> <tr> <td>active</td> <td></td> </tr> </tbody> </table>	<i>depolarisation</i>	<i>repolarisation</i>	closed	open	open			no diffusion	some diffusion out	rapid diffusion out	active		5	<b>Mark the first answer in each empty box.</b> If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = <b>0 marks</b>  <b>Award 1 mark per correct row</b>
<i>depolarisation</i>	<i>repolarisation</i>																
closed	open																
open																	
	no diffusion																
some diffusion out	rapid diffusion out																
active																	



Question		Answer	Marks	Guidance
	(c) (i)	sodium channels cannot open / AW ;  <i>idea that</i> opening depends on (correct) voltage / charge voltage too low for them to open ;  sodium ions/Na <sup>+</sup> , cannot, diffuse in / AW ;  no gradient for Na <sup>+</sup> ions ;	2	<b>DO NOT CREDIT</b> if given as a list with potassium channels <b>ACCEPT</b> 'sodium channels are closed'  <b>ACCEPT</b> 'because they are voltage-gated channels'  <b>DO NOT CREDIT</b> 'Na' but penalise once only
	(ii)	<i>idea that</i> keeps action potential moving in one direction ;  <i>idea that</i> action potentials (only) possible in next node ;  <i>idea that</i> action potentials not possible in , node behind active node / AW ;  <i>idea that</i> limits the frequency of nerve impulses ;	2	<b>LOOK FOR</b> <i>idea that</i> action potentials are only possible in the section ahead of the zone.
	(d)	<i>idea that</i> refractory period is diastole ; gives time for (chambers) filling ; (so) every systole followed by a diastole ;  AVP ;	2	<b>ACCEPT</b> 'time for heart to fill'. <b>LOOK FOR</b> <i>idea that</i> a period of contraction is followed by a period of relaxation.  e.g. prevents tetanic contraction of cardiac muscle / described.
	(e) (i)	nerve conduction velocity test / NCV test ;	1	<b>CREDIT</b> a description e.g. timing how long it takes for a muscle to contract when a nerve is stimulated. <b>OR</b> applying a stimulus and timing how long it takes to register sensation

Question		Answer	Marks	Guidance
	(ii)	(formation of) scar tissue / glial scars / AW ;  (presence of (axon) growth inhibitors / named growth inhibitor ;  axons have weak regeneration response ;  blood leaks into site of damage ;	2	<b>ACCEPT</b> ref to activity of glial cells or astrocytes  e.g. fibrinogen , CSPGs, (Chondroitin sulfate proteoglycans), proteoglycans, neurocan, brevican, phosphacan, versican  <b>ACCEPT</b> idea that cell division is prevented to prevent overgrowth  <b>ACCEPT</b> idea that blood clots form at damaged site
		<b>Total</b>	<b>16</b>	

Question		Answer	Marks	Guidance										
3	(a) (i)	(in) nucleus ;  (on) <u>X</u> chromosome ;	2	<b>ACCEPT</b> on a sex chromosome <b>DO NOT CREDIT</b> on the sex chromosomes / X or Y										
	(ii)	modifies protein into glycoprotein ;  idea that encloses Factor VIII into secretory vesicles ;	2	<b>ACCEPT</b> idea of adding sugars to protein <b>or</b> glycosylation  <b>ACCEPT</b> idea of packaging for <u>exocytosis</u> or for transport in vesicles out of the cell.  <b>DO NOT CREDIT</b> 'processes and packages' without further explanation										
	(b) (i)	<table border="1" style="margin-left: auto; margin-right: auto;"> <thead> <tr> <th>number of individual</th> <th>genotype</th> </tr> </thead> <tbody> <tr> <td>3 and 8</td> <td><math>X^H X^H</math></td> </tr> <tr> <td>4</td> <td><math>X^h Y</math> ;</td> </tr> <tr> <td>6</td> <td><math>X^H X^h / X^h X^H</math> ;</td> </tr> <tr> <td>7</td> <td><math>X^H Y</math> ;</td> </tr> </tbody> </table>	number of individual	genotype	3 and 8	$X^H X^H$	4	$X^h Y$ ;	6	$X^H X^h / X^h X^H$ ;	7	$X^H Y$ ;	3	<b>CREDIT</b> Y° for 'Y'
number of individual	genotype													
3 and 8	$X^H X^H$													
4	$X^h Y$ ;													
6	$X^H X^h / X^h X^H$ ;													
7	$X^H Y$ ;													

Question	Answer	Mark	Guidance
	<p>(ii) probability = 50% / 0.5 / ½ ;</p> <p><b>plus two from</b></p> <p>(or) can (also) be, heterozygous / <math>X^H X^h</math> / described ;</p> <p>(because) 2 / mother, is, heterozygous / a carrier / <math>X^H X^h</math> ;</p> <p>inherit either <math>X^H</math> or <math>X^h</math> from, individual 2 / mother ;</p> <p>can (only) inherit <math>X^H</math> from, individual 1 / father ;</p>	<p>1</p> <p>2</p>	<p><b>CREDIT</b> 1 mark for probability and any two for explanation</p> <p><b>IGNORE</b> ref to <math>X^H X^H</math> as genotype of 3 and 8 has been given in previous question</p> <p><b>CREDIT</b> explanations from a genetic diagram <b>OR</b> pedigree</p> 

Question		Answer	Mark	Guidance
	(c) (i)	<p><i>similar to type 1</i> MIDD is, treated with insulin injections / insulin dependent ; people are not obese / BMI less than 30 ;</p> <p><i>similar to type 2</i> MIDD is , late / mature , onset ;</p>	3	<b>ACCEPT</b> ref to statement 'develops around age of 30'
	(ii)	<p><i>inherited by both males and females</i> mitochondria / organelles, present in (2<sup>o</sup>) oocyte ; <i>idea that</i> oocyte will be fertilised by either X-carrying sperm or Y-carrying sperm (to produce males and females) ;</p> <p><i>cannot be passed on by males</i> no, mitochondria / organelles, (other than nucleus) come from sperm cell (at fertilisation) ;</p>	2	Idea that only nucleus from sperm cell contributes to zygote
		<b>Total</b>	<b>15</b>	

Question			Answer	Marks	Guidance
4	(a)	(i)	A iris ; B lens ; C anterior chamber / <u>aqueous</u> humour ;	3	
		(ii)	Idea that function of cornea is to refract light ;  <i>Idea that</i> light rays not converging correctly onto the lens ;  (light rays) not converging onto / AW, retina, properly <b>OR</b> light rays not hitting, fovea / macula ;	2	<b>ACCEPT</b> idea of light (rays) bending <b>DO NOT CREDIT</b> reference to reflection or diffraction  <b>DO NOT ACCEPT</b> reference to reduced amounts of light on retina
	(b)		7.3 ;;	2	<b>Correct answer = 2 marks</b>  If answer is not correct, <b>award</b> 1 mark for working $((2\ 711 - 2\ 512) \div 2\ 711) \times 100$ <b>OR</b> $(199 \div 2711) \times 100$  If answer not given to 1 decimal place, <b>award</b> 1 mark for seeing working as above <b>or</b> correct unrounded answer (ie 7.3404647....)
			<b>Total</b>	<b>7</b>	

Question		Answer	Marks	Guidance
5	(a)	maintaining a stable environment / AW ;  within tissue fluids / blood ;	2	<b>CREDIT</b> idea of parameters kept within set limits <b>IGNORE</b> constant  <b>IGNORE</b> reference to 'the body' without further qualification  <b>Note</b> – 'Maintaining a stable internal environment' = 2 marks

Question	Answer	Mark	Guidance
(b)	<p><i>For either hormone:</i>  (hormone released) into blood (plasma) ;  <i>ADH</i>  1. (stimulus is), low(er) water potential / too little water, in blood (plasma) ;  2.(change) detected by / AW, osmoreceptors , in hypothalamus ;  3. ADH released from (posterior) pituitary gland ;  4. (ADH) binds / AW, to cells / receptors, (in wall of) collecting duct ;  5. response is , (more) water reabsorbed (from collecting duct / <u>increase</u> in selective reabsorption of water ;  <i>thyroxine</i>  6. (stimulus is long term) fall in, (body/ AW), temperature ;  7. (change) detected by thermoreceptors in hypothalamus ;  8. hypothalamus stimulates (anterior) pituitary / described ;  9. thyroid stimulating hormone / <b>TSH</b> , released (from pituitary) ;  10. (TSH stimulates) thyroxine release from thyroid ;  11. (thyroxine stimulates), <u>increased</u> respiration rate / metabolic rate / <u>more</u> mitochondria formed ;  12. (response is) <u>more</u> heat energy released / AW ;</p>	9	<p><b>CREDIT REVERSE ARGUMENT</b> for ADH and a rise in water potential</p> <p>1. <b>CREDIT</b> description of osmoreceptor cells shrinking in response to low water (potential) <b>OR</b> that low water potential is the trigger.</p> <p><b>IGNORE</b> reference to receptors unqualified</p> <p>3. <b>DO NOT CREDIT</b> anterior pituitary</p> <p>4. <b>ACCEPT</b> <i>idea</i> that the collecting duct is the target organ for ADH</p> <p>5. <b>LOOK FOR IDEA</b> that increased water uptake is the <b>outcome</b></p> <p>6. <b>CREDIT</b> <i>idea</i> that fall in temperature is the trigger for thyroxine release</p> <p>7. <b>IGNORE</b> reference to receptors unqualified</p> <p>8. <b>CREDIT</b> description of <b>TRH</b> release from hypothalamus to pituitary</p> <p>9. <b>DO NOT CREDIT</b> TSH from posterior pituitary</p> <p>10. <b>CREDIT T3 and T4</b> for 'Thyroxine'</p> <p><b>ACCEPT</b> <i>idea</i> that respiration is <b>exothermic</b> and there is more respiration for mp <b>11</b> and <b>12</b></p>



Question	Answer	Mark	Guidance
	QWC ;	1	<b>Award if</b> the answer has been awarded mark points <b>1, 2 and 5</b> from ADH <b>AND</b> mark points <b>6, 7 and 11 or 12</b> from thyroxine

Question		Answer	Mark	Guidance
	(c) (i)	oedema / described ; proteins present in urine ; high creatinine levels in plasma ; reduced volume of urine ; AVP ; ;	2	<b>IGNORE</b> ref to glucose in urine.  e.g. skin rash metabolic acidosis reduction in EPO production
	(ii)	kidney stones ; high blood pressure ; (side effect of / AW) diabetes (mellitus) / high blood glucose ;  kidney infection ; kidney inflammation ; AVP ; ;	2	<b>any two from</b> e.g. polycystic kidney disease sudden blood loss chemical damage/drugs/alcohol physical trauma genetic predisposition
	(iii)	donation from someone who has died / AW ; living donor qualified / AW ;  non-human source / xenotransplantation ;	2	<b>Mark the first two suggestions</b> <b>CREDIT</b> example e.g. relative OR a living donor with close tissue match
	(d)	urine, is very dilute/has high water potential / low specific gravity ; water moves into cells, by osmosis/down a water potential gradient ; red blood cells, burst/haemolyse ;	2	
<b>Total</b>			<b>20</b>	

Question		Answer	Marks	Guidance
6	(a)	CTAT ;	1	<b>IGNORE</b> repeats of CTAT
	(b)	(i) to control / AW , the pH ; to stop the polymerase denaturing / to optimise pH for enzyme activity ;	2	<b>ACCEPT</b> reference to 'enzymes' (as assuming that they are referring to polymerase) <b>IGNORE</b> ref to proteins
		(ii) <i>Ideas that</i> it is a source of energy/AW ;	1	The key idea here is to make the link between the phosphorylated deoxynucleotide and provision of / release of energy.
	(c)	(i) <i>idea of</i> many hydrogen bonds holding the strands together ; hydrogen bonds break ; (because of) increased, kinetic energy / vibrations ;	2	
		(ii) (stabilised by) bonds between R groups ; strong / covalent, bonds ; disulfide, bonds / bridges ; AVP ;	2	<b>IGNORE</b> ref to Taq polymerase being heat-stable (as given in the question)  e.g. Taq polymerase obtained from bacteria living in hot springs <b>OR</b> Ref to Taq having a higher optimum temperature
	(d)	(i) (because) more hydrogen bonds (with GCGC) ;  GC pairs have 3 (hydrogen) bonds <b>AND</b> AT pairs have 2 (hydrogen) bonds ;	2	'A and T have 2 hydrogen bonds whereas C and G have 3' gets <b>both</b> marks.

Question		Answer	Marks	Guidance
	(iii)	<p><i>heading (a)</i> temperature ; °C ;</p> <p><i>heading (b)</i> <u>mean</u> (% GCGC sample of DNA denatured) ;</p>	3	<p><b>IGNORE</b> 'average' <b>ACCEPT</b> 'mean average' <b>DO NOT CREDIT</b> mean time <b>OR</b> mean temperature as these are contradictions</p>
(e)	(i)	<p><u>electrophoresis</u> ;</p> <p><b><i>plus a maximum of 4 marks from</i></b></p> <p>load samples into wells in (agarose) gel ;</p> <p>add, electrolyte / AW, solution / buffer ;</p> <p>connect electrodes/AW ;</p> <p>DNA samples migrate (from negative) to positive, electrode / end ;</p> <p>due to negatively charged phosphate (groups on DNA) ;</p> <p>smaller pieces move, further/ faster/ ORA ;</p> <p>AVP ;</p>	5	<p>Credit <b>1</b> mark for the name and up to <b>4</b> marks for the technique.</p> <p><b>ACCEPT</b> phonetic spelling</p> <p><b>ACCEPT</b> distance moved is inversely proportional to the size.</p> <p>e.g. detail of loading (such as addition of glycerol) use of , a tracking dye/bromophenol blue</p> <p><b>IGNORE</b> reference to visualising bands with stains</p>

Question			Answer	Marks	Guidance									
		(ii)	3 ; ;	2	<table border="1"> <tr> <td></td> <td>8</td> <td>12</td> </tr> <tr> <td>15</td> <td>(band at) 15 and (band at) 8</td> <td>(band at) 15 and (band at) 12</td> </tr> <tr> <td>8</td> <td>(band at) 8 (and 8)</td> <td>(band at) 8 and (band at )12</td> </tr> </table> <p><b>Correct answer = 2 marks</b> If the answer is incorrect, <b>ALLOW</b> 1 mark for correctly identifying <u>all</u> the possible combinations (as shown above).</p> <p><b>CREDIT</b> 3 out of 4 (3/4)</p>		8	12	15	(band at) 15 and (band at) 8	(band at) 15 and (band at) 12	8	(band at) 8 (and 8)	(band at) 8 and (band at )12
	8	12												
15	(band at) 15 and (band at) 8	(band at) 15 and (band at) 12												
8	(band at) 8 (and 8)	(band at) 8 and (band at )12												
			<b>Total</b>	<b>22</b>										

Question			Answer	Marks	Guidance
7	(a)	(i)	X beta/ $\beta$ , amyloid protein ; Y tau protein ;	2	<b>Mark the first answer on each prompt line.</b> If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = <b>0 marks</b>
		(ii)	smoking ; obesity ; high (saturated) fat diet ; lack of exercise ; high salt diet ; high blood pressure ; stress ; high blood cholesterol ; AVP ; ;	2	<b>Mark the first answer on each prompt line.</b> If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = <b>0 marks</b>  <b>any two from</b> e.g. atherosclerosis diabetes CHD stroke old age genetic predisposition qualified e.g. Familial Hypercholesterolaemia traumatic head injury
		(iii)	<i>AD</i> <i>idea that tau / amyloid , proteins build up over time ;</i>  <i>VD</i> damage/AW, may only affect one (small) area (at a time) <b>OR</b> repeat formation of blood clots;	2	<b>ACCEPT</b> idea that a part of the brain is deprived of oxygen
	(b)		MRI / fMRI ( scan) ; PET (scan) ; CT scan / CAT scan ;	2	<b>Mark the first answer on each prompt line.</b> If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = <b>0 marks</b>
<b>Total</b>				<b>8</b>	

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