

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
BOD	benefit of the doubt
NBOD	benefit of the doubt not given
ECF	error carried forward
	information omitted
I	ignore
R	reject

Question		Answer	Marks	Guidance
1	(a)	<p><i>combined HRT</i> oestrogen taken continuously ; progesterone taken for, a few / (10–14), days (each month) ;</p> <p><i>continuous</i> both hormones, without a break / every day ;</p>	3	<p>ACCEPT idea that progesterone taken for part of the cycle</p> <p>ACCEPT idea that 'HRT' is taken without a break as the names of the two hormones are given in the question.</p>
	(b)	a <u>placebo</u> was used ;	1	
	(c)	<p><i>Endometrial cancer:</i></p> <p>1 no significant difference OR idea that only slightly decreased risk with HRT ;</p> <p><i>HRT advantage</i></p> <p>2 idea that reduces risk of colorectal cancer AND hip fractures ;</p> <p>3 data in support (of any single factor) ;</p> <p><i>HRT disadvantage</i></p> <p>4 increases risk of CHD AND strokes AND breast cancer ;</p> <p>5 data in support (of any single factor) ;</p>	4	<p>DO NOT CREDIT ref to fewer cases with HRT unqualified</p> <p>comparing HRT with placebo, including correct use of units x in 10 000 women <u>per year</u> Either two values of x or a calculated difference LOOK FOR correct units given at least ONCE</p>
		<p>QWC ;</p> <p><i>DATA FOR 3 and 5</i></p>	1	<p>comparing HRT with placebo, including correct use of units (x in 10 000 women per year if unit quoted previously)</p> <p>Award if mark points 3 <u>and</u> 5 have been awarded</p> <p>SEE FIGURES on following page</p>

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

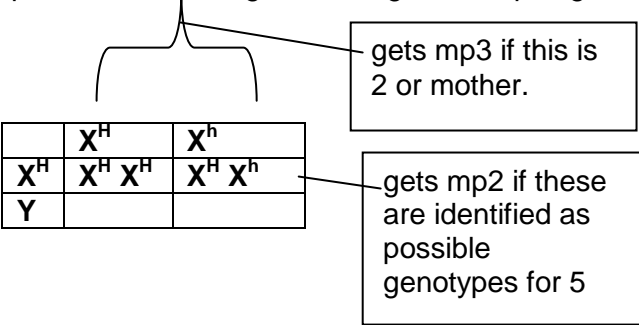
Question		Answer	Marks	Guidance
	(d)	<p><i>hysterectomy</i> (hysterectomy is) removal of, uterus / womb ;</p> <p>would not be able to investigate endometrial cancer ;</p> <p><i>blood pressure</i> hypertension / high blood pressure, (also) increases risk of, CHD / strokes ;</p> <p><i>for either</i> this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

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 and out (of axon) ; presence of, sodium - potassium pumps ;	1	CREDIT Na ⁺ and K ⁺ for 'ions' throughout CREDIT reference to <u>exchange</u> of ions IGNORE 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	Mark the first answer in each empty box. If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = 0 marks Award 1 mark per correct row
<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 ⁺ , cannot, diffuse in / AW ; no gradient for Na ⁺ ions ;	2	DO NOT CREDIT if given as a list with potassium channels ACCEPT 'sodium channels are closed' ACCEPT 'because they are voltage-gated channels' DO NOT CREDIT '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	LOOK FOR <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	ACCEPT 'time for heart to fill'. LOOK FOR <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	CREDIT a description e.g. timing how long it takes for a muscle to contract when a nerve is stimulated. OR 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	ACCEPT ref to activity of glial cells or astrocytes e.g. fibrinogen , CSPGs, (Chondroitin sulfate proteoglycans), proteoglycans, neurocan, brevican, phosphacan, versican ACCEPT idea that cell division is prevented to prevent overgrowth ACCEPT idea that blood clots form at damaged site
		Total	16	

Question		Answer	Marks	Guidance										
3	(a) (i)	(in) nucleus ; (on) <u>X</u> chromosome ;	2	ACCEPT on a sex chromosome DO NOT CREDIT on the sex chromosomes / X or Y										
	(ii)	modifies protein into glycoprotein ; idea that encloses Factor VIII into secretory vesicles ;	2	ACCEPT idea of adding sugars to protein or glycosylation ACCEPT idea of packaging for <u>exocytosis</u> or for transport in vesicles out of the cell. DO NOT CREDIT '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>$X^H X^H$</td> </tr> <tr> <td>4</td> <td>$X^h Y$;</td> </tr> <tr> <td>6</td> <td>$X^H X^h / X^h X^H$;</td> </tr> <tr> <td>7</td> <td>$X^H Y$;</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	CREDIT Y^o 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 / $\frac{1}{2}$; plus two from (or) can (also) be, heterozygous / $X^H X^h$ / described ; (because) 2 / mother, is, heterozygous / a carrier / $X^H X^h$; inherit either X^H or X^h from, individual 2 / mother ; can (only) inherit X^H from, individual 1 / father ;</p>	<p>1 2</p>	<p>CREDIT 1 mark for probability and any two for explanation</p> <p>IGNORE ref to $X^H X^H$ as genotype of 3 and 8 has been given in previous question</p> <p>CREDIT explanations from a genetic diagram OR pedigree</p> <div style="text-align: center;">  </div> <p>gets mp3 if this is 2 or mother.</p> <p>gets mp2 if these are identified as possible genotypes for 5</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	ACCEPT ref to statement 'develops around age of 30'
	(ii)	<p><i>inherited by both males and females</i> mitochondria / organelles, present in (2^o) 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
		Total	15	

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 OR light rays not hitting, fovea / macula ;	2	ACCEPT idea of light (rays) bending DO NOT CREDIT reference to reflection or diffraction DO NOT ACCEPT reference to reduced amounts of light on retina
	(b)		7.3 ;;	2	Correct answer = 2 marks If answer is not correct, award 1 mark for working $((2\ 711 - 2\ 512) \div 2\ 711) \times 100$ OR $(199 \div 2711) \times 100$ If answer not given to 1 decimal place, award 1 mark for seeing working as above or correct unrounded answer (ie 7.3404647....)
			Total	7	

Question		Answer	Marks	Guidance
5	(a)	maintaining a stable environment / AW ; within tissue fluids / blood ;	2	CREDIT idea of parameters kept within set limits IGNORE constant IGNORE reference to 'the body' without further qualification Note – '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 / TSH , 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>CREDIT REVERSE ARGUMENT for ADH and a rise in water potential</p> <p>1. CREDIT description of osmoreceptor cells shrinking in response to low water (potential) OR that low water potential is the trigger.</p> <p>IGNORE reference to receptors unqualified</p> <p>3. DO NOT CREDIT anterior pituitary</p> <p>4. ACCEPT <i>idea</i> that the collecting duct is the target organ for ADH</p> <p>5. LOOK FOR IDEA that increased water uptake is the outcome</p> <p>6. CREDIT <i>idea</i> that fall in temperature is the trigger for thyroxine release</p> <p>7. IGNORE reference to receptors unqualified</p> <p>8. CREDIT description of TRH release from hypothalamus to pituitary</p> <p>9. DO NOT CREDIT TSH from posterior pituitary</p> <p>10. CREDIT T3 and T4 for 'Thyroxine'</p> <p>ACCEPT <i>idea</i> that respiration is exothermic and there is more respiration for mp 11 and 12</p>

Question	Answer	Mark	Guidance
	QWC ;	1	Award if the answer has been awarded mark points 1, 2 and 5 from ADH AND mark points 6, 7 and 11 or 12 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	IGNORE 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	any two from 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	Mark the first two suggestions CREDIT 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	
Total			20	

Question		Answer	Marks	Guidance
6	(a)	CTAT ;	1	IGNORE repeats of CTAT
	(b) (i)	to control / AW , the pH ; to stop the polymerase denaturing / to optimise pH for enzyme activity ;	2	ACCEPT reference to 'enzymes' (as assuming that they are referring to polymerase) IGNORE 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	IGNORE ref to Taq polymerase being heat-stable (as given in the question) e.g. Taq polymerase obtained from bacteria living in hot springs OR Ref to Taq having a higher optimum temperature
	(d) (i)	(because) more hydrogen bonds (with GCGC) ; GC pairs have 3 (hydrogen) bonds AND AT pairs have 2 (hydrogen) bonds ;	2	'A and T have 2 hydrogen bonds whereas C and G have 3' gets both marks.
	(ii)	number of repeats ; the longer the DNA/the more repeats, the greater the number of hydrogen bonds ; OR length of time heated for ; more , vibrations/kinetic energy, as time increased ;	2	Mark the first suggestion. DO NOT CREDIT any references to temperature as this is the independent variable.

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>IGNORE 'average' ACCEPT 'mean average' DO NOT CREDIT mean time OR mean temperature as these are contradictions</p>
(e)	(i)	<p><u>electrophoresis</u> ;</p> <p><i>plus a maximum of 4 marks from</i></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 1 mark for the name and up to 4 marks for the technique.</p> <p>ACCEPT phonetic spelling</p> <p>ACCEPT 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>IGNORE 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>Correct answer = 2 marks If the answer is incorrect, ALLOW 1 mark for correctly identifying all the possible combinations (as shown above).</p> <p>CREDIT 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												
			Total	22										

Question			Answer	Marks	Guidance
7	(a)	(i)	X beta/ β , amyloid protein ; Y tau protein ;	2	Mark the first answer on each prompt line. If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = 0 marks
		(ii)	smoking ; obesity ; high (saturated) fat diet ; lack of exercise ; high salt diet ; high blood pressure ; stress ; high blood cholesterol ; AVP ; ;	2	Mark the first answer on each prompt line. If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = 0 marks any two from 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) OR repeat formation of blood clots;	2	ACCEPT idea that a part of the brain is deprived of oxygen
	(b)		MRI / fMRI (scan) ; PET (scan) ; CT scan / CAT scan ;	2	Mark the first answer on each prompt line. If the answer is correct and an additional answer is given that is incorrect or contradicts the correct answer then = 0 marks
Total				8	

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