19. Genetic Technology

19.2 Genetic Technology Applied to Medicine

Paper 4

Marking Scheme

Q1.

(a)	1 family history of, breast / ovarian, cancer ; *plus any **three** from:* 2 (mutation causes) change of primary structure (of tumour suppressor protein) ; 3 changes, folding / bonding / 3D shape / tertiary structure (of tumour suppressor protein) ; 4 non-functional (tumour suppressor) protein formed ; 5 DNA is not repaired / cells with damaged DNA do not die ; 6 (so) increases number of mutations / mutations accumulate ; 7 (some mutations) cause uncontrollable, cell division / mitosis (of breast cells) ;	**4**

(c)	6.1×10^4 ;; **A** 6.125×10^4 *for two marks* *allow one mark for* 61 250 **or** $\frac{3.5 \times 10^7}{400} = 8.75 \times 10^4$ or 87 500	**2**

(d)	*any **two** from:* *supports:* 1 (women with *BCRA1*) have a higher <u>probability</u> of survival ; **ora** 2 (for women with *BRCA1*) the probability of survival decreases less **or** there is a smaller rate of decrease of probability of survival **or** bigger difference between the two groups over time for the probability of survival ; **ora** 3 data quote to show differences between the survival of the two groups at a particular time ;	**2**

time / months	probability of survival	
	with mutation	without mutation
30	0.975 range 0.975 – 0.988	0.85
60	0.95	0.75
90	0.85	0.675 range 0.675 – 0.688
120	0.75	0.63 range 0.613 – 0.625
150	0.75	0.55
180	0.7 / 0.70 range 0.7 – 0.71	0.475 / 0.474 / 0.47 range 0.46 – 0.475

4 study carried out for, a long time / 15 years ;

does not support:
5 no statistical test carried out / *ref. to* sample size ;

Q2.

(b)(i)	*any **three** from:* 1 **add**, healthy / normal / correct / functional, gene / allele, to virus ; 2 inject virus into eye ; 3 gene integrated into (eye cell) genome / DNA ; 4 gene / allele, expressed to produce functioning, protein / enzyme ; 5 AVP ; e.g. adeno-associated virus / AAV voretigene neparvovec (drug)	**3**
(b)(ii)	*any **one** from:* 1 easily accessible / easy to inject ; 2 low risk of immune response ; 3 (eye small in size so) small amount of gene therapy treatment needed / AW ;	**1**

(c)(i)	*any **three** from:* 1 (overall) improvement in acuity of both eyes **or** (overall visual) acuity of both eyes becomes more negative ; 2 treated eye, has **more** improved acuity / has **more** negative (visual) acuity ; **ora** 3 *ref. to* anomalous result at month 9 / result that does not fit the pattern at month 9 ; 4 data quote – values of one eye at two months **or** values of both eyes at one month ;	**3**

time / months	change in visual acuity / au	
	eye not treated	eye treated
0	0.00	0.00
1	-0.12	-0.20
2	+0.02	-0.40
3	0.00	-0.50
4	-0.13	-0.52
5	-0.22 or -0.23	-0.52
6	-0.15	-0.54
7	-0.12	-0.50
9	-0.15	-0.33
10	-0.08	-0.52
12	-0.12	-0.54

Q3.

(a)(ii)	*any **three** from:* *(protein made in, bacteria / yeast, so)* **1** can be made in, large / unlimited, quantities ; **2** cheap to make / low cost / cost-efficient ; **3** does not, harm / involve, animals / pigs / cows ; **4** no risk of (named) disease, transfer / infection ; *(sequence identical to human sequence so)* **5** does not cause, allergy / inflammatory response / immune response / antibody production ; **6** acts more rapidly ; **7** do not, develop tolerance to it / need larger doses ;	**3**
(b)(i)	cow ;	**1**

Q4.

	*any **seven** from:* 1 genetic engineering / recombinant (DNA technology) ; 2 (named) drug / treatment / protein, from, (GM) bacteria / (GM) yeast / (named) (GM)O ; 3 make insulin (drug for diabetes) ; 4 make factor VIII (drug for haemophilia) ; 5 make adenosine deaminase (drug for SCID) ; 6 genetic, screening / diagnosis / testing ; 7 (detect) BRCA1 / BRCA2 ; 8 (detect number of repeats / allele for) Huntington's ; 9 (detect allele for) cystic fibrosis / CFTR ; 10 example of action if diagnosis positive ; 11 gene therapy ; 12 insert / add, normal allele into (named), cells / tissue (of person) ; 13 to treat, eye disease(s) / LCA / Leber's congenital amaurosis ; 14 to treat SCID ; 15/16 AVP ; ; e.g. *ref. to* help / develop, xenotransplantation GM / antigen-free, pigs / animals, to source, tissues / organs, for transplants GM plants to make vaccines GM / recombinant / DNA / mRNA, vaccines recombinant antibody technology other / different, recombinant therapeutic protein / genetic screening / gene therapy, example	**7**

Q5.

(b)(i)	1 stem cells, proliferate / divide / multiply / make new cells ; 2 (correct) gene is, inherited by / passed on to / present in, new cells ;	**2**

(b)(iii)	*any **two** from:* 1 retrovirus / new gene, could / must not, insert in wrong place / disrupt other genes ; 2 could / must not, cause cancer ; 3 could / must not, cause, infection / disease ; 4 could / must not, cause, immune / allergic, response ; 5 AVP ;	**2**

Q6.

(a)	*diabetes:* insulin from (GM) bacteria ; **A** ref to, yeast / animal cells *single gene disorder:* gene therapy **or** insert, functional / normal / dominant, allele into, target / somatic / stem, cells **or** genome ; **A** gene **R** *replacing one allele with another*	**2**

Q7.

(a)	*any **three** from:* 1 repeat treatment causes, bigger / more rapid, immune response ; 2 (so) target cells (expressing new protein) short-lived / AW ; 3 decrease in (trans)gene expression on re-administration ; 4 AVP ; e.g. *ref. to* antibody binding / antibodies attract phagocytes	**3**
(b)	*any **four** from:* *naked DNA* 1 has to be injected into target cell / lack of organ-specific delivery ; 2 low efficiency of cellular uptake ; 3 rapidly broken down ; *viruses* 4 small packaging capacity / only small amount of DNA can be carried ; 5 low probability of integration (into host genome) ; 6 cause mutations in host DNA / (gene) insertion disrupts gene function / insertional mutagenesis ; *liposomes* 7 low ability to, add DNA / genes, into target cells (genome) / low transduction efficiency ;	**4**

Q8.

(c)	*any **two** from:* *arguments for* 1 more diagnosis, means better management of those with the deficiency ; 2 can prevent losing further children to the deficiency / can decide to not have children / can choose to have children after embryo testing ; 3 *ref. to* possibility of using enucleated donor egg for mutated mtDNA ; *any **two** from:* *arguments against* 4 different opportunities for people to access the test ; 5 no cure ; 6 some people person may still lead a normal life ; 7 many mutations still unknown so may still not lead to positive diagnosis ;	**3**

Q9.

(b)(i)	*any **two** from:* *if child has, sickle cell anaemia / SCT* **1** informed decision can be made about continuing with the pregnancy / AW ; **2** treatment can begin quickly (after birth) ; **3** parents can prepare, mentally / emotionally / for the cost of treatment ; **4** (this) child can make an informed decision about having children when older ; *if child does not have, sickle cell anaemia / SCT* **5** removes worry / AW ;	**2**

Q10.

(c)	*advantage:* **1** treat (genetic) disease / treat people / reduce suffering / therapeutic ; *problem (max 1):* **2** possible side effects (on human) / drug may not be pure / may contain allergens / risk of harm / may be unsafe / risk of goat disease transmission to humans ; **3** process may harm goats / animal welfare issues ; **4** vegetarians / vegans, may not take drug ;	**2**

Q11.

(a)	*any **four** from:* 1 identical to human, insulin / protein ; **R** similar 2 no, allergic reaction / immune response / side effects ; 3 human protein may, have high(er) activity / work better / (more) rapid response ; 4 no chance of developing tolerance (to animal insulin) ; 5 no risk of transmitting diseases ; 6 supply unlimited / large-scale production ; **ora** not enough animal pancreases to meet demand 7 cost (of production) low(er) ; 8 no ethical objections / no religious objections ; **ora**	**4**

Q12.

(a)(i)	*any 2 of:* 1 to treat disease caused by, faulty / recessive, allele ; **A** cure 2 deliver, gene / allele / DNA (into target cells of individuals) ; 3 named example ; e.g. SCID / cystic fibrosis	**2**
(a)(ii)	(reverse transcriptase) uses mRNA to make (ss)DNA ; (DNA polymerase) makes DNA double-stranded / description ;	**2**
(a)(iii)	1 to prevent virus spreading (throughout the body) ; 2 to limit, side effects / immune response / cancer / illness / infection / cell destruction ;	**2**
(b)(i)	*any 3 of:* 1 control ; 2 *idea of* to see if gene therapy has worked **or** to compare the effect of no gene therapy with gene therapy **or** to compare the effect of no hG-CSF gene with presence of hG-CSF gene ; 3 to see where the gene (carried by the vector), goes (in, body / brain) **or** to see if the gene (carried by the vector), enters cells **or** to see where cells have been transformed ; 4 to see the effects of the, vector / virus, alone ;	**3**

(b)(ii)	*any 1 of:* 1 (control) to compare results (of mice without stroke) to mice with stroke ; 2 to establish baseline figures (in mice without a stroke) / act as a reference point ; 3 to make the study valid ;	**1**

(c)	*any 4 of:* *survival* 1 increases survival ; *brain damage* 2 reduces (percentage of brain occupied by) fluid filled space ; *behaviour test* 3 improves / lowers, behavioural test score ; **A** score closer to normal *general* 4 comparative data quote **to support**, mp1 / mp2 / mp3 ; **A** processed data mp1: 25% v 63%; 25% v 100%; 63% v 100% mp2: 5.2% v 3.6%; 5.2% v 3.0%; 3.6% v 3.0% mp3: 0.90 v 0.67 5 eye drops four times is better than eye drops once ; **ora** 6 if effective with mice likely to be effective with humans ; 7 *ref. to* unknown differences between mice and humans / study is only on mice ; 8 treatment is, non-invasive / quick ;	**4**