

Inherited Disorders 2

QUESTION 1

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)	(genotype / gametes from T / mother) h and h (accept h) (genotype / gametes from U / father) H and h offspring genotypes: correctly derived from correct gametes chance = 50% / ½ / 0.5 / 1 : 1 / 1 out of 2	eg may be in punnett square allow own upper and lower case symbols or allow any symbol correctly used with key do not accept 1 : 2 allow irrespective of working	1 1 1 1
b)i)	(cells from) embryo tested for particular allele / gene / (named) genetic condition / the disease / gender	ignore x-rays / ultra sound	1
b)ii)	any one from: ☑ possible harm to embryo or mother ☑ sensible reference to abortion ☑ hard to make moral / ethical decisions	ignore moral / ethical unqualified ignore not natural / playing God references to other ideas eg designer babies must be well argued	1
c)i)	(cystic fibrosis is) recessive (allele / gene) need two alleles / genes or need one allele / gene from each parent or both parents must have cystic fibrosis allele / gene	allow not dominant	1 1
c)ii)	any three from: ☑ osmosis / diffusion ☑ more concentrated solution outside cells / in mucus ☑ water moves from dilute to more concentrated solution ☑ partially permeable membrane (of cell)	do not accept reference to movement of ions / solution by osmosis / diffusion assume concentration is concentration of solute unless they tell us otherwise allow correct references to movement of water in relation to concentration	3

		gradient	
c)iii)	ribosomes		1
Total marks			12

QUESTION 2

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)i)	circle unshaded	mark independently could be in body of script	1 1
a)ii)	(Harriet) dd DD Dd	in first box if another letter is chosen it must be used throughout and upper or lower case must be clear	1 1 1
b)i)	to check for the D allele.		1
b)ii)	any one from: ☒☒☒ may harm / kill foetus / embryo / baby / mother ☒☒☒ immoral / unethical / religion	allow could affect the baby ignore playing God ignore references to unnatural ignore wrong unqualified ignore expense / prejudice unqualified ignore lack of permission ignore results are unreliable	1
Total marks			7

QUESTION 3

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)i)	recessive allele		1
a)ii)	carriers		1
b)i)	6		1
b)ii)	1 in 4 / 0.25 / ¼ / 25 % / 1:3	do not accept '3:1' / 1:4 / 1 in 3 /	1
c)	advantage: detect CF qualified – eg at early stage / before becoming pregnant or (only) healthy children produced disadvantage: some embryos are destroyed / may damage embryo		1 1
Total marks			6

QUESTION 4

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)	changes code /sequences of bases or sequence of amino acids is different the enzyme has different / wrong shape / structure so substrate will not fit into enzyme / will not join to enzyme	allow the active site is changed	1 1 1
b)i)	46	allow 23 pairs	1
b)ii)	also inherited (from mother) normal chromosome 15 / normal allele / normal gene / boy is heterozygous / Hh (allele for) this disorder is recessive or the normal allele would give a working enzyme	allow the boy is a carrier ignore converse	1 1
b)iii)	genetic diagram including: Parental gametes: H and h from both parents derivation of offspring genotypes: HH Hh Hh hh identification of hh (having the disorder) if 1 in 4	accept alternative symbols, if defined allow alternative if correct for student's parental genotypes / gametes	1 1 1
Total marks			9

QUESTION 5

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)i)	1		1
a)ii)	fertilisation / fusion	allow sexual reproduction allow fertilise / fuse ignore joining	1
b)i)	Dd		1
b)ii)	dd		1
c)i)	1 in 2		1
c)ii)	1 in 2		1
Total marks			6

QUESTION 6

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)i)	one form of a / one gene	do not allow 'a type of gene' allow a mutation of a gene	1
a)ii)	not expressed if dominant / other allele is present / if heterozygous or only expressed if dominant allele not present / or no other allele present	allow need two copies to be expressed / not expressed if only one copy / only expressed if homozygous	1
b)i)	two parents without PKU produce a child with PKU / 6 and 7 \square 10	allow 'it skips a generation'	1
b)ii)	genetic diagram including: Parental gametes: 6: N and n and 7: N and n derivation of offspring genotypes: NN Nn Nn nn identification: NN and Nn as non-PKU OR nn as PKU correct probability only: 0.25 / $\frac{1}{4}$ / 1 in 4 / 25% / 1 : 3	accept alternative symbols if defined allow genotypes correctly derived from student's parental gametes allow correct identification of student's offspring genotypes do not allow 3 : 1 / 1 : 4 do not allow if extra incorrect probabilities given	1 1 1 1
c)i)	mitosis	correct spelling only	1
c)ii)	8		1
c)iii)	DNA	allow deoxyribonucleic acid do not allow RNA / ribonucleic acid	1
d)i)	may lead to damage to embryo / may destroy embryos / embryo cannot give consent	allow avoid abortion allow emotive terms – eg murder religious argument must be qualified allow ref to miscarriage allow idea of avoiding prejudice against disabled people allow idea of not producing designer babies	1
d)ii)	any one from: • prevent having child with the	ignore ref to having a healthy child	1

	disorder / prevent future suffering / reduce incidence of the disease <ul style="list-style-type: none"> embryo cells could be used in stem cell treatment 	ignore ref to selection of gender allow ref to long term cost of treating a child (with a disorder) allow ref to time for parents to become prepared	
Total marks			12

QUESTION 7

QUESTION	ANSWER	EXTRA INFORMATION	MARKS
a)i)	female) has XX / only X's / no Y	allow has X chromosomes ignore ref to genes / cells	1
a)ii)	extra chromosome / has 47 chromosomes / one set has 3 copies no. 18	ignore reference to chromosome numbers other than 47 or no. 18	1 1
b)i)	14	allow in range of 13.5 to 14.5	1
b)ii)	7	allow in range of 6.75 to 7.25 accept ecf from 5bi	1
c)	Advantages: any two from: <ul style="list-style-type: none"> more than 1 embryo (so more chance of success) tested at 3 days cf 10 weeks or tested earlier tested before pregnancy no termination / abortion spare embryos have a potential use Disadvantages: any one from: <ul style="list-style-type: none"> needs an operation (spare) embryos / human life destroyed / harmed higher cost embryos might not implant / might not develop 	allow method 2 may cause a miscarriage tested when only 3 days old accept described hazard of operation must be comparative	2 1
Total marks			8