QUESTIONSHEET 1

(a) gene mutations alter the base sequence on DNA thus altering the genetic code;
chromosomal mutations alter the chromosome structure thus altering the sequences of genes on the chromosome;
chromosome mutations may also alter the chromosome number of the individual; **3**

(b) substitution/deletion/addition/inversion/translocation;;(any two)
sickle cell anaemia/albinoism/melanism/any other valid example;;(any two) **4**

(c) when an individual receives three copies of chromosome 21 instead of two;
reference to non disjunction/translocation;
reference to learning difficulties/thick set bodies/thick necks/infertility/any other correct symptom; **max 2**

(d) one gene mutation will produce a new character which will thus be a discontinuous variant;
thousands of similar gene mutations will form a wide range of slightly different characters which will give continuous variation; **2**

TOTAL 11

QUESTIONSHEET 2

(a) (i) increase in chromosome number;
either of individual chromosomes or of complete sets; **2**

(ii) allopolyploidy; **1**

(iii) the genomes of one set of (the species') chromosomes; **1**

(iv) where the chromosomes fail to separate during anaphase;
due to failure of the spindle to contract;
thus new nuclear membrane forms round both lots of chromosomes together; **max 2**

(b) the individuals arising from polyploidy can all interbreed to form fertile offspring (and thus are a species);
they cannot breed back to the parental stock to produce fertile offspring and are thus separate species to the parents;
due to the impossibility of chromosomes to pair/form bivalents in meiosis; **3**

(c) (i) they possess the genes of both parental species including the genes that confer the best/strongest characters;
thus have a 'double dose' of valuable genes;
giving them greater survival potential; **3**

(ii) increased yield;
easier to thresh/extract grain/ better flour/higher disease resistance/quicker maturing/better nutritive value; **2**

TOTAL 14

QUESTIONSHEET 3

(a) mosquitoes were exposed to DDT to eradicate them as malarial vectors;
alleles mutated in a few mosquitoes and gave them DDT resistance;
these resistant forms survived and reproduced to form more resistant mosquitoes;
because non-resistant forms had been wiped out there was no competition;
and resistant forms could flourish/less selection pressure on resistant forms; **max 4**

(b) over use of antibiotics by medical profession/in animal foodstuffs exposed many bacteria to antibiotics;
mutant alleles appeared which gave certain strains of bacteria antibiotic resistance;
the mutant alleles were in the plasmid DNA;
when bacteria die the plasmids are released into the substrate;
and may become incorporated into other species of bacteria (thus giving them antibiotic resistance); **max 4**

(c) due to the formation of a restitution nucleus/equivalent, that doubles the chromosome number;
thus it can now form bivalents in meiosis and so produce gametes; **2**

TOTAL 10

QUESTIONSHEET 4

(a) radiation that is naturally present in the environment;
comes from cosmic rays hitting Earth/ from radioactive elements (such as uranium/thorium/radon) in the Earth's rocks /from natural radioactive carbon and potassium isotopes in biological matter; **2**

(b) alpha = helium nuclei; beta = electrons/positrons; gamma = electromagnetic energy;
beta is least likely since its particles are light and have little penetrating power; **4**

(c) mustard gas; 5-bromouracil/dioxin/any other valid example; **2**

(d) (i) there is a linear relationship between the dose of radiation over a total life span and the amount of mutation /the higher the dose the more mutation and time between doses does not limit the mutation; **1**

(ii) $X = \frac{12.5 \times 2}{5}$; = 5%; **2**

(e) (i) Any three of: substitution/addition/deletion/inversion/translocation; **1**

(ii) Any three of: deletion/addition/inversion/translocation; **1**

TOTAL 13

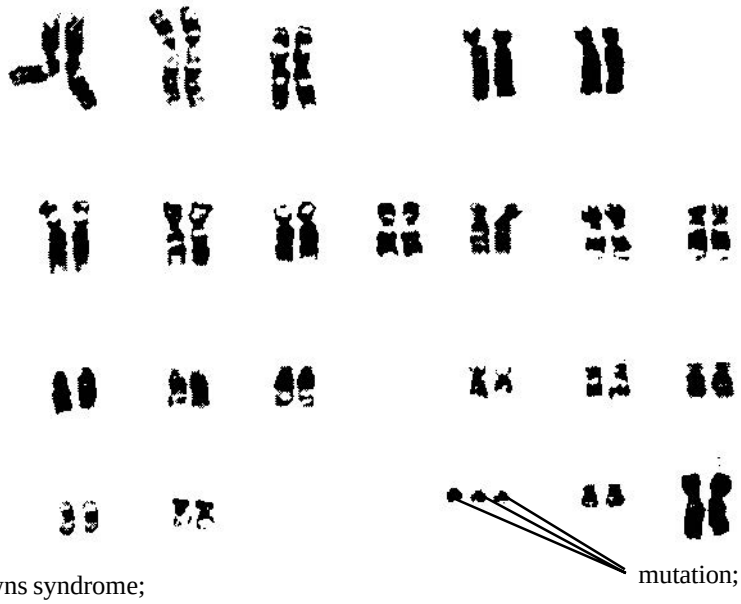
QUESTIONSHEET 5

(a) (i) cells (in culture) treated with colchicine/drug;
which holds chromosomes in metaphase of mitosis/in their most visible state;
cells smeared on slide, (fixed) and stained to show chromosomes;
chromosomes in many nuclei photographed through microscope;
chromosomes cut out singly and matched into pairs;
according to length/shape/position of centromere/staining pattern; **max 4**

(ii) amniotic fluid sample taken from pregnant mother;
this contains fetal cells/cells shed from the baby;
these can be gently centrifuged to concentrate them;
then placed in tissue culture;
allowed to grow and divide (mitotically) for several days; **max 3**

(b) (i) yes;
46 plus 1 extra chromosome/22 pairs plus a group of 3 identical chromosomes; **2**

(ii)



trisomy 21/Downs syndrome;
ref to non disjunction;
chromosomes 21 failed to separate in anaphase and both went to same pole/egg nucleus;
thus when egg was fertilised the zygote contained three of chromosome 21; **max 4**

(iii) female;
if it was male a different shaped Y chromosome would be visible; **2**

TOTAL 15

QUESTIONSHEET 6

(a) (i) DNA triplet would become CAT;
this would form GUA by transcription to the mRNA;
GUA codes for valine;
thus sixth amino acid in chain/penultimate amino acid is changed from glutamic acid to valine; **max 3**

(ii) will affect/alter the cross-bonding in the globin chain/polypeptide;
which will alter the 3-D shape/conformation/tertiary structure of the molecule; **2**

(iii) substitution; **1**

(b) malarial parasite develops inside red cells of humans;
cannot survive on haemoglobin S as substrate/cannot survive in reduced potassium ion environments;
thus sickle cell sufferers are resistant to malarial infection but usually die from sickle cell anaemia at an early age;
sickle cell trait heterozygotes are resistant to malarial infection and do not die from sickle cell anaemia;
thus reproduce normally raising incidence of mutant gene in the population of the malarial zone;
(probably) have a greater reproductive capacity than malarial sufferers (within the population); **max 4**

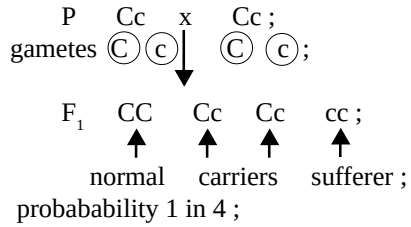
TOTAL 10

QUESTIONSHEET 7

(a) (i) (three) nucleotides could be omitted during replication of DNA (in meiosis/gamete formation);
thus mRNA does not include the omitted nucleotides (during transcription);
thus an amino acid (actually phenylalanine) will be omitted from the polypeptide chain;
during translation;
thus CFTR protein will not work/work properly/be ineffective; **4**

(ii) substitution; insertion/addition; translocation; **max 2**

(b) (i) (use suitable symbols, eg C for normal allele, c for cystic fibrosis allele)



probabability 1 in 4 ; **5**

(ii) CF males are sterile so their genes can be disregarded;
4% of the population are carriers;
thus chances of carriers crossing = .04 x .04 = .0016 or 0.16%; (allow other ways of showing figures)
probability of carrier cross producing a CF child is 1 in 4;
thus expected incidence will be $\frac{0.16}{4}$ = 0.04%;
(if say $\frac{100}{2500}$ = 0.04% allow 1 mark only, unless explained) **max 4**

(iii) because new mutations (of the same type) are constantly happening; **1**

(c) remove mucus by physiotherapy/thoracic massage/aspiration;
diet control (to counteract pancreatic misfunction);
gene therapy; **max 2**

TOTAL 18

QUESTIONSHEET 8

(a) (i) ref to restitution nucleus;
failure of chromosomes to separate during anaphase;
most likely in mitosis in apical meristem;
thus this part/sector of the plant would be tetraploid and would produce diploid gametes;
since inbreeding these would produce tetraploid seed/offspring;
which would breed on to produce more tetraploids; **max 4**

(ii) gametes of Coffea arabica will contain 22 chromosomes;
gametes of other species/ancestral form will contain 11 chromosomes;
thus accurate bivalent formation in meiosis cannot occur;
so even if hybrid grows it will not be able to produce viable gametes; **max 3**

(b) genes from other stock will not be incorporated and so it becomes genetically stable;
all plants will have basically the same genotype;
good features/high yield/disease resistance/flavour/quick growth/any valid example, will be perpetuated/ not diluted by intrusion of other genes;
polyploidy will be maintained;
polyploids produce larger beans; **max 4**

TOTAL 11

QUESTIONSHEET 9

(a) pollen of the one species fertilises the ovule/embryo sac of the other species;
seeds produced;
these germinate/grow to produce offspring with one set of chromosomes from each of the parent species;
ref to F_1 hybrid;
ref to allopolyploidy; **max 3**

(b) no meiosis can occur;
since (non-matching) chromosomes of orange and pummelo will not pair (in synapsis);
thus haploid pollen/egg nuclei cannot be produced;
seed develops purely by mitosis; **max 2**

(c) no variation due to meiosis/random assortment/chiasmata;
no variation due to outbreeding/fertilisation;
variation can only occur by mutation; **max 2**

(d) gene mutation/point mutation (of a gene); (reject 'polyploidy)
ref to substitution/deletion/addition/inversion/translocation (of genes); **2**

TOTAL 9

QUESTIONSHEET 10

(a) change in the base sequence of the gene;
due to substitution/deletion/addition/inversion/translocation of bases;
alters the codon sequence of the gene;
(thus) alters/may alter the amino acid sequence of the polypeptide made by the gene; **max 3**

(b) (i) sickle cell anaemia;
haemophilia;
(red-green) colour blindness; (allow other correct examples) **max 2**

(ii) non-disjunction;
sets of chromosomes fail to segregate correctly to poles (in anaphase of meiosis);
results in egg cell containing two of chromosome 21;
when fertilised results in trisomy 21/Down's syndrome baby has three copies of chromosome 21;
another cause of Down's syndrome is due to translocation of part of another chromosome onto the end of chromosome 21;
max 3

TOTAL 8

QUESTIONSHEET 11

(a) (i) incidence remains very low/less than 2 per thousand/does not increase up the the age of 30 (years);
steady increase/increases to 4 per thousand/incidence doubles between 30 and 35 (years);
steeper increase/increases to 18 per thousand from 35 to 45 (years); **3**

(ii) meiosis becomes less efficient as mother ages/random assortment/segregation to poles less efficient;
older parents have undergone longer exposure to possible mutagens and so tend to have higher mutation rates; **max 1**

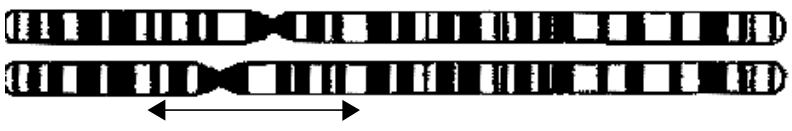
(iii) non-disjunction;
failure of sets of chromosomes to segregate accurately (in anaphase of meiosis);
for instance two of chromosome 21/18/13 may go to one pole and none to the other pole;
(thus) egg may contain two copies of the chromosome;
(thus) after fertilisation the zygote will have three copies of the chromosome; **max 3**

(b) ref amniocentesis;
collect amniotic fluid (which contains fetal cells);
centrifuge to collect fetal cells and then grow them in tissue culture;
treat with colchicine/drug to hold chromosomes in metaphase/visible/spread out state;
make smears and stain (with chromosome stain);
photograph chromosomes of many nuclei, cut out and pair/ref karyotyping; **max 3**

TOTAL 10

QUESTIONSHEET 12

(a) (i) inversion; **1**

(ii) **A**
B

1

(b) (i) cross bandings show positions of stained/similar DNA/DNA bands marked by gene probes;
cross bandings match very closely (in non-mutated part of chromosomes);
even match in mutated region taking into account the inversion;
since they have similar staining/marked DNA/similar DNA distribution they are probably closely related; **max 3**

(ii) after divergence from common stock/common ancestry; **1**

TOTAL 6