

- T-killer cells seek out the antigen and destroy it or enhance the activity of macrophages which will then destroy it.
- (b) Injections (vaccines) may result in either artificial passive immunity or artificial active immunity.
 - In artificial passive immunity, antibodies are injected into the blood stream. [1]
 - This form of protection only lasts as long as the antibodies last. [1]
 - In artificial active immunity, the weakened (attenuated); dead antigen or inactivated toxin (toxoid) is injected so a person manufactures their own antibodies without developing the disease. [1] They provide much longer protection against diseases. [1]
- (c) Quarantine – isolation of infected individuals; close country borders in a pandemic, wash hands with soap and water, dry properly; cover mouth when coughing or sneezing; put used tissues in a rubbish bin; do not share drink bottles or lip glosses; avoid sharing needles; use safe-sex practices. [3]

TT 6 – MUTATIONS

Section 1: Multiple Choice (30 marks)

- | | |
|------|-------|
| 1. d | 6. c |
| 2. d | 7. b |
| 3. c | 8. b |
| 4. b | 9. c |
| 5. b | 10. d |

Section 2: Short Answer (50 marks)

- Mutant
 - Pedigree
 - Congenital
 - Allele
 - Somatic cell
 - Deleterious
 - Phenotype
 - Autosome (autosomal chromosome)
 - Meiosis
 - Cancer

[10]
- Mutations are spontaneous random changes [1] in the nucleotide sequence of DNA. [1]
 - Viruses such as rubella, HPV, chemicals such as benzene, formaldehyde, radiation such as X-rays, ultraviolet (UV), gamma rays. [3 groups and 3 examples = 6]
 - These can produce mutations because they could interfere with the replication of DNA [1], alter the sequence of bases in a DNA strand [1], cause parts of chromosomes to move position or invert. [1]
- Somatic cell mutation. This is a change to

a gene or a chromosome in a body cell. [1] The consequences of such a change may be insignificant OR serious depending how it affects the cell. [1] As these occur in a body cell, they are not passed on to offspring. [1] Somatic cell mutations are the cause of cancer.

Germ-line (or gametic) mutation. This is a change to a gene or a chromosome in a gamete or zygote [1] and therefore can be passed on to the offspring and possibly to their offspring in turn. [1] e.g. phenyl ketonuria (PKU). Therefore, it can have serious consequences. [1]

- Environmental influences can affect the rate of mutations in a population. [1] However, the environment cannot cause a particular mutation to occur. [1]
 - A mutant gene may be transmitted through many generations unchanged. [1] It may mutate again producing another unusual feature or it may revert back to the original gene. Because it may persist for many generations it is possible for natural selection to act on it. [1]
- Useful mutations are very rare. [1] Random change to a gene which is functioning well is more likely to lead to its dysfunction rather than improvement. [1]
- Advantageous mutations are most important. [1] They offer increased chances of survival and are selected by the environment. They are responsible for a wide variety of well adapted organisms which have appeared on Earth. [1]
 - Unfavourable mutations are selected against. [1] They are not likely to persist in a population for long. Natural selection works against them and eventually removes them from the population. [1]
- Klinefelter's Syndrome. [1]
 - Male. [1]
 - Forty-seven chromosomes. [1]
- Female. [1]
 - An extra twenty-first chromosome. [1]
 - Down syndrome (trisomy 21). [1]
 - Non-disjunction (when the chromosomes fail to separate) of homologous chromosomes during either anaphase I or anaphase II of meiosis. [2]
- Degeneracy refers to the fact that a number of codons determine the same amino acid. [2] E.g. AAG and AAA both code for the amino acid lysine [any example, 1]. Therefore the mutation of changing the G to an A does not change the amino acid [1] and therefore does not change the protein. [1]

Section 3: Extended Answer (20 marks)

[Each bullet point = 1 mark]

(a)

- Mutations may arise through random chance or through environmental influence.
- Environmental factors include certain chemicals (e.g. benzene), some viruses, X-rays, ionising radiation and UV radiation.
- Individuals need to be educated to avoid excess or unnecessary exposure to these factors (diet, smoking, alcohol, drugs).
- These agents cause a change to the DNA which can cause cancer or inherited defects.
- A change to the DNA in somatic cells can cause cancer which is not inherited.
- A change to the DNA in a germ-line (gametic) cell may be inherited.
- Most people carry many defective, recessive genes that are hidden by dominant, normal genes.
- Only the homozygous recessive genotype will affect the phenotype.
- The homozygous recessive phenotype is more likely to appear in closely related marriages, e.g. between first cousins.

(b)

- Researching (and drawing) a family pedigree, genetic counsellors may be able to determine the probability of a couple having a child with a particular genetic disease.
- E.g. if the couple are determined by the pedigree to be heterozygous for a particular disease, the likelihood of any child having the disease would be 0.25.
- Phenyl ketonuria (PKU), cystic fibrosis and sickle-cell anaemia are diseases which can be identified in this way.
- If the probability of having a child with a serious genetic disease is unacceptably high, the couple may choose to adopt or use assisted reproduction technology (e.g. IVF, GIFT, etc.), combined with donor gametes/s if necessary.
- Genetic testing provides a profile of an individual's DNA though it is not entirely complete.
- Genetic testing can show chromosomal abnormalities or the presence of abnormal proteins (which are indicators of abnormal genes).
- These tests can be carried out on both the parents and foetus (prenatal).
- Prenatal testing may involve amniocentesis, umbilical blood sampling or chorionic biopsy to examine the karyotype of the developing child.
- Karyotypes can be used to test for a number of conditions, including Down's syndrome.
- Karyotypes can be used to determine the sex of the foetus and therefore in the case

of sex-linked diseases, help to determine the probabilities of inheritance.

- Infant screening for metabolic factors may detect genetic diseases such as Phenyl ketonuria (PKU), which can be treated and in this case, cured, if detected early.

17 – GENE POOLS

Section 1: Multiple Choice (30 marks)

- | | |
|------|-------|
| 1. b | 6. a |
| 2. b | 7. a |
| 3. b | 8. d |
| 4. c | 9. c |
| 5. d | 10. d |

Section 2: Short Answer (50 marks)

- (i) pedigree
- (ii) evolution
- (iii) population
- (iv) genotype
- (v) independent assortment
- (vi) migration
- (vii) natural selection
- (viii) crossing over
- (ix) mutation
- (x) genetic biodiversity
- (xi) adaptation
- (xii) speciation

[12]

2.

- (a) True
- (b) False
- (c) False
- (d) True
- (e) True
- (f) False
- (g) True
- (h) True
- (i) True

[9]

3.

- (a) Gene frequency refers to the percentage of members of a population with a particular allele. [2]
- (b) Isolation reduces gene frequencies in populations. [1] Gene flow increases gene frequencies. [1]
- (c) Isolation is caused by parts of a population being cut off from other parts of the population so gene flow is reduced. [1] Barriers to gene flow may be geographical (increase in sea level, mountain ranges) or cultural. [1]

Gene flow refers to the movement of genes from one population to another as a result of interbreeding between members of the two different populations. [1] This introduces new variations into the population, altering