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 Phenylketonuria (PKU), which can be treated and in this case, cured, if detected early.*

TT 7 – GENE POOLS

Section 1: Multiple Choice (30 marks)

1.	*b*	6.	*a*
2.	*b*	7.	*a*
3.	*b*	8.	*d*
4.	*a*	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, alterin

the frequency of genes. [1]

4.

(a) *The Founder effect refers to the situation in which a sample of a population moves into or migrates into a new area. The resulting population reflects the sample of genes from the original 'founding' population. [1] E.g. the Dunkers in America, or Pitcairn Islanders who are the descendants of sailors from the ship,* HMS Bounty *and a group of Tahitian people. [1]*

(b) *The Founder effect reduces the variety of genes in gene pools as the population is established by a sample of individuals from the original population and they only carry a sample of the genes from the original population. [1]*

(c) *The bottleneck effect occurs when a disaster happens and the original population is just reduced to a few survivors. [1] The range and the frequency of alleles will change. [1]*

(d) *If a volcano erupts on a small island and most of the population die as a result, or invasion and genocide by an invading force. [1]*

5.

(a) *These people may be isolated geographically and tend to interbreed with each other due to cultural and religious beliefs. [2]*

(b) *By a blood test. [1]*

(c) *Gene therapy in which the defective gene is replaced by a normal working one. [2]*

6. *Immigration refers to the movement of individuals into an area [1] while emigration refers to individuals leaving an area. [1]*

7.

(a) *Tay-Sachs is due to a mutation that affects a metabolic pathway. It could be an example of founder effect because it occurs in the Ashkenazim at much higher frequency [1] than in the rest of the population (1 in 3600 compared to 1 in 40 000 births). Presumably, the mutation occurred in some Ashkenazim people [1] and because of their intermarriage, stayed within their people [1] even if they migrated around the world.*

(b) *Natural selection favours those individuals best suited to the environment at the time. [1] Sufferers of Tay-Sachs would die before reproductive age so would not pass on the gene [1] so one would expect the incidence to decrease over time, [1] particularly if heterozygotes within the community knew the risks, had genetic testing and made the decision not to have children.*

(c) *If Tay-Sachs confers a survival advantage for tuberculosis, it is an advantage for heterozygotes to carry the gene. [1] If they survive better in TB prone areas, they can then pass the gene onto their offspring. [1]*

Section 3: Extended Answer (20 marks)

(a)

- *Natural populations tend to produce lots of offspring.*
- *Variation occurs between members of a population.*
- *All members of a population compete for food, shelter, mates, etc.*
- *Some organisms within the population are better suited to the environment than others.*
- *This is due to the variations which make them more suited to the environment*
- *The more suitable organisms survive and reproduce and pass their favourable features onto their offspring.*
- *The organisms with unsuitable or unfavourable features die out.*
- *Organisms with suitable or favourable features gradually increase in number in the population.*
- *This is 'survival of the fittest' (to the environment).*

[2 marks per point = 16]

(b)

- *Over time the most favourable features become more common in the population.*
- *The genes responsible for the favourable features increase in frequency.*
- *Variation in the population decreases.*
- *In other words, natural selection reduces gene frequency in a population. [4]*

TT 8A – BIOTECHNOLOGY

Section 1: Multiple Choice (30 marks)

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

Section 2: Short Answer (50 marks)

1.

(a) *Biotechnology is the use of micro- organisms and their biological processes for human benefit. More recently it involves the manipulation of micro-organisms to fulfil a human demand or need. [2]*

(b) *It is used in brewing (wine and beer) and baking, making yoghurt and cheese as well as the manufacture of hormones such as insulin and Factor VIII used in blood clotting. [any 2 = 1]*

2.

(i) *Polymerase chain reaction (PCR).*
(ii) *Electrophoresis.*
(iii) *DNA profiling.*
(iv) *Plasmid.*
(v) *Clone.*