In this section each statement is followed by five related statements, only ONE of which is correct. Put [T] in front of the correct answer.

1. Chromosomal disorders: -

(a) Include Klienfelter's syndrome, which occurs only in the females. [ ]

(b) Result from single point mutation. [ ]

(c) Include Turner syndrome which is 45, X. [ ]

(d) Occur at a frequency less than single gene disorders. [ ]

(e) In trisomy 18, the numbers of chromosomes are 2n-1. [ ]

2. Multifactorial disorders:

(a) Tend to concentrate in families. [ ]

(b) Follow Mendelian inheritance. [ ]

(c) Include Down's syndrome. [ ]

(d) Are single gene disorders. [ ]

(e) Always occur in adult life. [ ]

3. Ahmad has a genotype TT, while his wife has a genotype tt (where T is dominant for tall height):

(a) All their children will be short. [ ]

(b) 50% of their children will be tall. [ ]

Cc) All their sons will be tall but daughters will be short. [ ]

(d) 50% of their children will have genotype TT. [ ]

(e) All their daughters and sons will be tall. [ ]

4. If a wife has a hemoglobin genotype AC and the husband has a hemoglobin genotype AS, then:

(a) There is 25% chance of having a child with Hb SS (sickle cell anaemia) [ ]

(b) There is 25% chance that the children will be carriers for C and 25% will

be carrier for S. [ ]

(c) There is 50% chance that the children will be double heterozygous for

Hb SC. [ ]

(d) All children will be severely anaemic. [ ]

(e) All children will have hypochromic microcytic anaemia. [ ]

5. Pleiotropy occurs frequently for autosomal dominant disorders. This means that:

(a) The disorder occurs in every generation. [ ]

(b) Unaffected persons will not have affected children. [ ]

(c) New mutations may occur. [ ]

(d) There are multiple phenotypic effects of the single genotype. [ ]

(e) The phenotype is normal, though the genotype is abnormal. [ ]

6. Pseudodominance:

(a) Refers to dominant disorders which are not expressed. [ ]

(b) Is shown by an autosomal recessive disorder if a homozygous marries a heterozygous for the same disorder. [ ]

(c) Is shown by blood group O. [ ]

(d) Is expressed in heterozygotes. [ ]

(e) Refers to reduced penetrance of a dominant disorder. [ ]

7. Human population is very diverse. This

(a) Diversity results as people from different countries have different genes at a loci in their genome. [ ]

(b) Is due to inheritance of different alleles of genes. [ ]

(c) Is due to presence of different metabolic pathways in their body. [ ]

(d) Is an effect of only the environmental factors. [ ]

(e) Is an effect of different foods that they eat. [ ]

8. If a mutation occurred in the intron of a gene, it will:

(a) Produce a protein with different amino acid sequence. [ ]

(b) Produce a variant protein with normal properties. [ ]

(c) Produce a variant protein with altered properties. [ ]

(d) Not be inheritable. [ ]

(e) Have no effect on the structure of the gene product. [ ]

9. Polymorphism:

(a) Refers to linked genes. [ ]

(b) Is shown by only DNA. [ ]

(c) Is shown by a gene that has no alleles. [ ]

(d) Refers to alleles that are common in a population. [ ]

(e) Is shown by sickle cell gene (Hb S) in central province of Saudi Arabia as 1 in 1000 people have this gene. [ ]

10. Alleles:

(a) Are same form of different genes. [ ]

(b) Are multiple forms of a gene that occupy different loci. [ ]

(c) Are always polymorphic. [ ]

(d) Are two or more forms of the gene that occupy the same locus. [ ]

(e) Do not segregate during meiosis. [ ]

11. Protein polymorphism is studied by:

(a) Using restriction endonucleases. [ ]

(b) DNA sequencing. [ ]

(c) Observation of variations in physical features. [ ]

(d) Using a microscope to determine chromosomal changes. [ ]

(e) Electrophoresis. [ ]

12. You suspected that your patient has thalassaemia as:

(a) His red cells were sickling. [ ]

(b) He had iron deficiency. [ ]

(c) He had hypochromic-microcytic anaemia with normal iron level. [ ]

(d) He had altered activity of G-6-PD. [ ]

(e) He had no anaemia. [ ]

13. ABO blood groups show polymorphism, so:

(a) Their frequency is the same in all populations. [ ]

(b) They all show autosomal recessive inheritance. [ ]

(c) The rarest allele occurs at a frequency of more than 1% in a population. [ ]

(d) Blood groups A & B are codominant. [ ]

(e) Blood group O is recessive. [ ]

14. Genes are said to be linked, if:

(a) They are allelic genes. [ ]

(b) Always segregate during meiosis. [ ]

(c) Occur at two loci which are over 50 centimorgan apart. [ ]

(d) The genetic distance between them is O centimorgan. [ ]

(e)the Lod score is —2 or less. [ ]

15. Centi Morgan

(a) Refers to the distance between two chromosomes: [ ]

(b) Is about 1 million bp. [ ]

(c) Is the distance between two alleles of a gene. [ ]

(d) Value is I if the chance of recombination between loci is 1/100000. [ ]

(e) is equal to 1000 million bp. [ ]

16. Haemoglobin:

(a) Has genes on chromosomes 11 and 16 [ ]

(b) Bind oxygen to the globin chains [ ]

(c) Transport CO_2 from lungs to tissues [ ]

(d) Has four globin chains link to one heme group [ ]

(e) Is a simple protein [ ]

17. in gene of globin chains of haemoglobin:

(a) There are three introns and two exons. [ ]

(b) The ε and ϒ genes are present on chromosome 16. [ ]

(c) All genes have three exons and two introns. [ ]

(d) Each person has two a-genes, one is inherited from mother and one from father. [ ]

(e) Pseudogenes (ψ) are not expressed as they have a mutation in exons. [ ]

18. Haemoglobinopathies:

(a) Are diseases that can be acquired. [ ]

(b) Are caused by mosquito bite. [ ]

(c) Result from mutation in the G-6-PD gene on X chromosome. [ ]

(d) Occur at a high frequency in areas which have past or present history of malaria endemicity. [ ]

(e) Are X-linked disorders. [ ]

19. Malaria does not occur in Hb S carriers, as:

(a) Mosquito does not bite the carriers. [ ]

(b) Mosquito cannot multiply in the blood of carriers. [ ]

(c) Mosquito does not inject the malaria parasite into the carriers. [ ]

(d) Carriers can kill the mosquitoes more easily. [ ]

(e) Malaria parasite is destroyed in the red cells of carriers. [ ]

20. There are many types of haemoglobinopathies:

(a) They result from mutations in introns of globin genes. [ ]

(b) They may produce haemoglobin with increased oxygen affinity. [ ]

(c) They never produce haemoglobin with altered structure. [ ]

(d) They are produced by increased production of globin chains. [ ]

(e) They increase the stability of haemoglobin. [ ]

21. Methaemoglobin has:

(a) High oxygen affinity. [ ]

(b) Slightly decrease oxygen affinity. [ ]

(c) No oxygen binding ability. [ ]

(d) Has no heme. [ ]

(e) has iron in ferrous (Fe^{++}) state. [ ]

22. Unequal crossing over of chromosomes produces:

(a) Hb S. [ ]

(b) Hb C. [ ]

(c) Hb O-Arab. [ ]

(d) Hb Riyadh. [ ]

(e) Mb Lepore. [ ]

23. In ϫδβ-thalassaemia:

(a) α-chains are not present. [ ]

(b) α-chains are decreased. [ ]

(c) γ-chains are normal. [ ]

(d) β-chains are increased. [ ]

(e) Hb A, Hb A_2 and Hb F are decreased. [ ]

24. cDNA for α-globin chains:

(a) Has three exons and 2 introns. [ ]

(b) Has a promoter site on 5' end. [ ]

(c) Is spliced to make a normal RNA. [ ]

(d) Is synthesized using mRNA. [ ]

(e) Is the same as the ct-globin gene on the chromosomes. [ ]

Answer the following questions as true [T] or false [F]

1. Vectors are double stranded DNA molecular that can be removed or injected into cells.
2. Polymerase chain reaction (PCR) is a technique used for gene cloning.
3. Viral diseases such as hepatitis can be diagnosed using DNA technology.
4. Restriction endonucleases recognize and hydrolyse palindromic sequences.
5. Bacteriophage lambda is used as a probe during DNA technology.
6. Plasmids are nuclear DNA in bacteria.
7. Hemophilia cannot be diagnosed from clinical presentation of a patient.
8. Abmad and Sara are both normal and are cousins. They have an autosomal dominant disorder in the family, so you council them non-directively against maniage.
9. G-6-PD deficiency can be diagnosed by measuring enzyme activity.
10. A double heterozygous state occurs if a person is heterozygous for two alleles.
11. In hydrops faetalis the genotype is --/-- as no β-globin genes are present.
12. The α/β ratio is increased in patients with α-thalassaemia.

In this section, each statement is followed by four related statements that may be all correct, all false or a mixture of true and false. Put [T] in front of the correct answer and [F] in front of the false one.

1. In sickle cell haemoglobin:

(a) The codon number 6 is mutated from GAG to GTG. [ ]

(b) Haemolysis of red cell occurs in sickle cell anaemia as Hb S is more soluble in deoxygenated state. [ ]

(c) The haemoglobin S cannot bind and transport O_2. [ ]

(d) Only the f3-globin chains have a change in one amino acid, while α-chains are normal. [ ]

2. The following mutation occurred in f3-globin gene of haemoglobin: -

UAA—*UAC:

(a) It will result in premature termination of β-globin gene of haemoglobin. [ ]

(b) It will result in a longer β-globin chain. [ ]

(c) It will result in β-thalassaeemia. [ ]

(d) it will not produce any defect in the β-globin chain. [ ]

3. Hb-lepore:

(a) May be caused by single point mutation in β and δ globin genes. [ ]

(b) Cannot be inherited by the children from parents. [ ]

(c) Is an abnormal haemoglobin not found in normal people. [ ]

(d) May be due to gene deletion. [ ]

4. Linkage analysis is used for:

(a) Gene mapping. [ ]

(b) Carrier detection. [ ]

(c) Diagnosis of a genetic disease. [ ]

(d) Gene cloning. [ ]