

Q1:Chromosomal abnormalities are found in the following diseases except:

- A.Turner syndrome
 - B.Acute Lymphocytic lymphoma
 - C.Angelman syndrome
 - D.Color blindness
 - E.Patau syndrome
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Q2:Cyclin proteins are given this name because they :

- A.They cycle between activation and inhibition
 - B. They cycle between expression and degradation
 - C. They cycle between free form and CdK-bound
 - D.They cycle between nucleus and cytoplasm
 - E. They cycle between the different phases of the cell cycle
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*** Hadoo el as2elh bdha fhem*****

Q3: rasmih 3n el probe men

Q4: rasmih 3n SNPs (4 SNPs were analyzed for the association with a disease, what is true about data)

- A.All SNPs are associated with the disease
 - B.
-

Q5:Above an efficient annealing temperature to amplify a DNA segment, there will be a problem, it is:

- A.The DNA polymerase loses its activity
 - B.The primer cant hybridize to the DNA
 - C.The DNA template can't be denatured
 - D.The Nucleotides become unstable
 - E.The DNA template is degraded easily
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Q6:All the following are caused by nondisjunctional phenomenon except:

- A.Klinefelter's syndrome
 - B.Retinoblastoma
 - C.Down's syndrome
 - D.Patau's syndrome
 - E.Edward's syndrome
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Q7:Which of the following is not a chromosome instability disorder:

- A.Xeroderma pigmentosum
- B.DiGeorge Syndrome
- C.Ataxia telangiectasia
- D.Fanconi anemia
- E.Bloom syndrome

Q8: Which of the following techniques is of no value in diagnosis of neural tube defects:

- A. Amniocentesis
 - B. Chorion villus sampling
 - C. Maternal serum screening
 - D. Ultrasonography
 - E. Alpha fetoprotein in amniotic fluid
-

Q9: Which of the following is the most common mode of inheriting a disorder : (Not sure if the Q was like this)

- A. Chromosomal Abnormality
 - B. Autosomal dominant
 - C. Autosomal recessive
 - D. Multifactorial
 - E. X-Linked recessive
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Q10: Karyotype can be obtained from all the following except:

- A. Sperm
 - B. Peripheral blood bone marrow
 - C. Fibroblast from skin biopsy
 - D. Epithelial cells from (el Sayar kan hik)
 - E. Solid tumor biopsy
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Q11: Which of the following is not a member of organic acidemia:

- A. Methyl malonic Acidemia.
 - B. Propionic Acidemia.
 - C. Metachromatic leukodystrophy
 - D. Multiple carboxylase deficiency.
 - E. Ketothiolase deficiency .
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Q12: Which of the following is not true about Foundations of Heredity Science –Mendelian-:

- A. Variable traits are inherited
 - B. *Gene* – trait-specific unit of heredity
 - C. Alternative versions of a gene (*alleles*) determine the trait
 - D. dependent assortment
 - E. Each parent *transmits* an allele to the offspring
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Q13: Lysosomal storage diseases is a Single-Gene “Mendelian” Disorders caused by :

- A. Structural proteins
- B. Enzymes and inhibitors
- C. Receptors
- D. Cell growth regulation
- E. Transporters

Q14: Trait is:

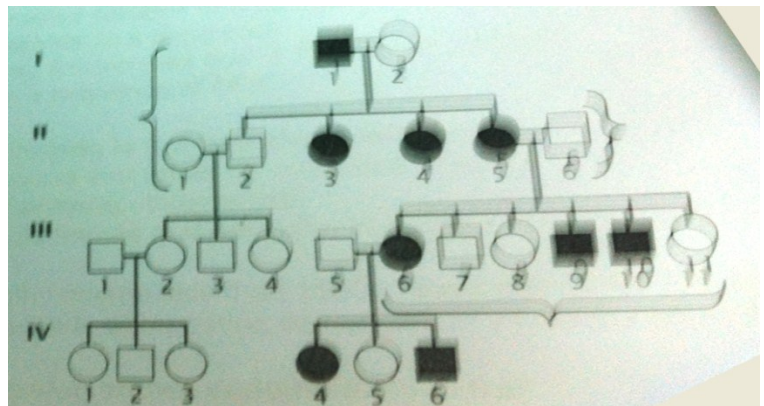
- A. a structure, function, or attribute determined by a gene or group of genes
 - B. the alternate forms of the character
 - C. the physical description of the character in an individual organism
 - D. the genetic constitution of the organism
-

Q15: Which of the following is not true about Mitochondrial inheritance:

- A. Matrilineal mode of inheritance: only mother passes mitochondrial DNA
 - B. Higher spontaneous mutations than nuclear DNA
 - C. Defects both male and female, but transmitted only through female
 - D. Differences of phenotypic severity is due to homoplasmy
 - E. Disorder involving mitochondrial DNA shows autosomal dominant pattern of inheritance
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Q16: What is the most probable mode of inheritance in this pedigree:

- A. Autosomal dominant
- B. Autosomal recessive
- C. X-linked dominant
- D. X-linked recessive
- E. Y-linked



Q17: Mutation disorder with several symptoms refers to:

- A. pleiotropy
 - B. Genetic heterogeneity
 - C. Variable expression
 - D. Germline mosaicism
 - E. Reduced penetrance
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Q18: Which of the following about X-linked Recessive Disorders is not true:

- A. Affects hemizygous males and homozygous females.
- B. Expressed phenotype much more common in males
- C. Affected males get the mutant allele from their mothers
- D. Typically associated with miscarriage or lethality in males.
- E. Daughters of affected males are usually heterozygous – thus unaffected

Q19: Family Pedigree usually summarize the family history of the patient, in order to obtain a pedigree which of the following question is not important:

- A. Ask whether relatives have a similar problem
 - B. Ask if there were siblings who have died
 - C. Inquire about miscarriages, neonatal deaths
 - D. Ask severity of the disease in the index formula
 - E. Ask about consanguinity
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Q20: Diabetes mellitus follow:

- A. Autosomal dominant
 - B. Autosomal recessive
 - C. X-linked
 - D. Mitochondrial
 - E. Multifactorial
-

Q21: Polyploidy Refers to:

- A. Extra copies of a gene adjacent to each other in a chromosome
 - B. An individual with complete extra set of a chromosome
 - C. A chromosome which has replicated but not inherited
 - D. Multiple ribosomes present on a single mRNA
 - E. An inversion which does not include the centromere
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Q22: The stage of meiosis in which chromosome pair and cross-over:

- A. Prophase I
 - B. Metaphase I
 - C. Prophase II
 - D. Metaphase II
 - E. Anaphase II
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Q23: Which of the following shows codominance is true:

- A. Has both alleles independently expressed in heterozygote
 - B. Has one allele dominant to the other
 - C. Has alleles tightly linked on the same chromosome
 - D. Has alleles expressed at the same time in the development
 - E. Has alleles that are recessive to each other
-

Q24: Which of the following is not true about telomerase:

- A. Seal chromosome and retain chromosome integrity
- B. Maintained by enzyme – telomerase
- C. Reduction in telomerase and decrease in number repeats important in ageing and death
- D. Telomere length became less than 1500 in base pairs after age of 65
- E. Telomeres are made of repeating sequences of TTAGGG on both strands

Q25: Which of the following is not true about genetic drift: (Sorry ☹)

- A. Fluctuation in the genetic frequencies
- B. The differences between allele frequencies between population which..... contact between them
- C. cut off one or more portions of a population causes a change in the frequency of alleles in a gene pool
- D. Change in allele frequencies due to functional mutation
- E. Any change in a sequence of DNA

Q26: Which of the following statement regarding polymorphism is wrong:

- A. SNPs occur every 300-1000 bases in human genome
- B. Non-coding SNPs can influence gene expression
- C. there are 5.6 million differences..... genome
- D. SNPs can occur in both coding and non-coding regions of the genome
- E. Non-synonymous: when single base substitutions do not cause a change in the resultant amino acid

Q27: Which of the following is true about retinoblastoma :

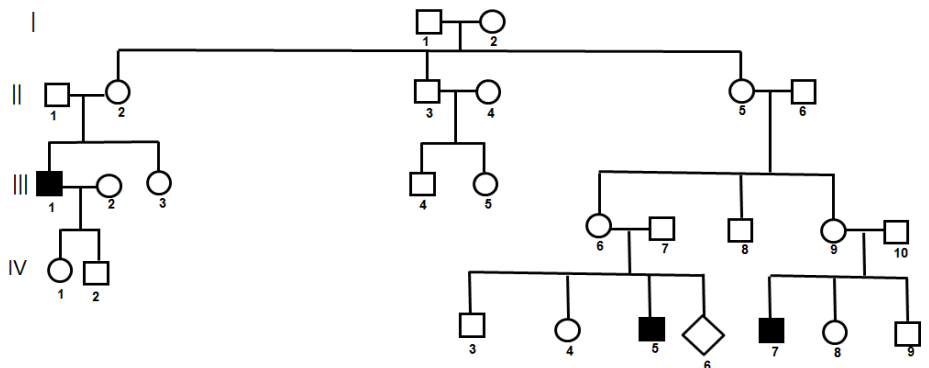
- A. Is associated with the loss of the short arm of chromosome 13
- B. Is caused by mutation in growth suppressor gene
- C. Is an autosomal dominant condition
- D. Is inherited in the majority of unilateral cases
- E. Is inherited in the majority of bilateral cases

Q28: The standard karyotype is performed by photomicrograph in which mitotic stage:

- A. Intraphase
- B. Prophase
- C. Metaphase
- D. Anaphase

Q29: Which female in the pedigree is carrier for the disease:

- A. I-2, II-4, III-5
- B. I-2, III-6, III-9
- C. III-3, IV-1
- D. II-5, III-3
- E. II-2, III-6, III-9, IV-1



Q30:A non-selective newborn screening program should contain all the following except:

- A.Minimal false positives
 - B.Minimal false negatives
 - C.Clearly defined disorder
 - D.well defined inheritance and pathogenesis
 - E.Advantage of early diagnosis
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Q31:Which of the following combination is not true:

- A.Primary ovary insufficiency/ Fragile mental retardation 1(FMR1)
 - B.Mitochondrial DNA/ largely associated with proteins
 - C.Osteogenesis imperfect/ mosaicism
 - D.Fragile X syndrome/ Hypermethylation of a CpG island
 - E.Uniparental disomy/ Absent of insulin in newborn
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Q32:Which of the following is not true regarding Urea Cycle Defects:

- A.Main function to prevent accumulation of N_2 waste as urea
 - B.High ammonia, low BUN
 - C.Possible lactic acidosis
 - D.Presence of ketonuria
 - E.Normal to mild low glucose
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