GENESIS

(Post Graduation Medical orientation Centre)

Exam : Genetics_Class Test_FCPS_2020

Class/Chapter :

Total Mark : 100
Pass Mark : 70

Question 26 to End is Based on Single Answers

Time : 5400 Min
Date : 2020-10-31

1. The following are autosomal dominant disorders
a). a) Neurofibromatosis
b). b) Polycystic kidney disease
c). c) Diabetes insipidus
d). d)Congenital adrenal hyperplasia
e). e) Achondroplasia
TTFFT

2. Autosomal recessive fearutes
a). a) Unaffected individual does not transmit the disease
b). b) Male –Female are equally affected
c). c) Complete penetration is common
d). d) Onset is usually late in life
e). e) Transmission pattern is oblique
FTTFF

3. X-linked disorders are
a). a) Myotonic dystrophy
b). b) Duchenne muscular dystrophy
c). c) Agammaglobulinemia
d). d) Lesch-Nyhan syndrome
e). e) Spinal muscular atrophy
FTTTF

4. The following statements are correct regarding X-linked recessive trait
a). a) Female are carriers due to random inactivation of one X chromosome
b). b) The gene responsible for the condition is transmitted from an affected man to all his daughters
c). c) The gene is not transmitted directly from father to son
d). d) A cannier female will transmit the condition to 25% of her sons
e). e) X-linked disorders are transmitted by heterozygous females to their son
TTTTT

5. Down's syndrome is
a). a) X-linked recessive disorder
b). b) Associated with increased maternal age
c). c) A trisomy 21 disorder
d). d) Associated with coarctation of aorta
e). e) Diagnosed by Barr body analysis
FTTFF

6. 4. A person with karyotype 47 xxy is associated with
a). a) Mongolism
b). b) A single barr body
c). c) Presence of an ovotestis
d). d) Gynaecomastia
e). e) Mental retardation
FTFTF

7. Turner syndrome is associated with
a). a) Primary amenorrhoea
b). b) 45XO karyotype
c). c) Rare mental retardation
d). d) Autosomal dominant disease
e). e) Male phenotype.
TTTFF

8. Robertsonian translocations
a). a) Are more common than reciprocal translocations
b). b) Will result in 45 chromosomes in the normal carrier
c). c) Are the commonest cause of down's syndrome
d). d) Involve only the submetacentric chromosomes
e). e) Result in 100% recurrence rate if both chromosomes 21 are involved (21/21 translocations)
FTFFT

9. The pedigree for autosomal dominant disorders demonstrates the following characteristics
a). a) De novo mutation
b). b) Complete penetrance
c). c) Variable expression
d). d) Vertical transmission
e). e) 50% recurrence risk in each pregnancy
TFTTT

10. Methods currently used to treat genetic diseases in humans include--
a). a) Germ-cell gene therapy
b). b) Stem-cell transplantation
c). c) Enzyme or protein replacement
d). d) Dietary restriction
e). e) In situ repair of mutations by cellular DNA repair mechanism
FTTTF

11. Examples of aneuploidies--
a). a) Down syndrome
b). b) Klinefelter syndrome
c). c) Turner syndrome
d). d) Fragile X syndrome
e). e) Pallister-killian syndrome
TTTFT

12. Regardingthe structure of DNA, the following information are correct--
a). a) DNA stands for deoxyribonucleic acid.
b). b) One complete DNA molecule consists of a single helix.
c). c) The building blocks of DNA are called nucleotides.
d). d) One nucleotide is composed of two parts: a sugar and a nitrogen base.
e). e) The sugar in DNA is called ribose.
TFTFF

13. Following diseases are Autosomal recessive
a). a) Wilson's disease
b). b) Thalassaemia
c). c) Polycystic kidney disease
d). d) Osteogenesis imperfect
e). e) Neurofibromatosis
TTFFF

14. Following statements are CORRECT regarding autosomal recessive disorders
a). a) Recurrence risk is 50%
b). b) H/O consanguinity is a must.
c). c) Incidence is more common than disorders of AD
d). d) Only homozygotes are affected.
e). e) No carrier state is seen.
FFFTF

15. Indications of prenatal diagnosis--
a). a) Women below 35 years of age.
b). b) Raised maternal serum α-fetoprotein.
c). c) Family H/O anencephaly
d). d) Family H/O down syndrome
e). e) Known carriers of phenylketonuria
FTTTT

16. Indication for Barr body
a). a) Unambiguous external genitalia
b). b) Lymphoedema in adult female
c). c) Inguinal mass in female
d). d) Cryptorchidism in male
e). e) Infertility in female
FFTTF

17. Disorders that show multifactorial inheritance-
a). a) Fallot's tetralogy
b). b) Multiple sclerosis
c). c) Glaucoma
d). d) Epilepsy
e). e) Bipolar mood disorders
TTTTT

18. Karyotyping procedure includes-
a). a) Isolation of an anucleated cell
b). b) Arrest of cell cycle at anaphase by adding colchicine
c). c) Re-attachment of the dividing cells
d). d) Fixation of cell by methanol
e). e) Micro-photography
FFFTT

19. Examples of genomic imprinting-
a). a) Angelman syndrome
b). b) Russell silver syndrome
c). c) Prader willi syndrome
d). d) Beckwith wideman syndrome
e). e) Down syndrome
TTTTF

20. Fragile X syndrome-
a). a) Most common cause of genetic mental retardation
b). b) Micrognathia
c). c) Squint
d). d) FMR1 gene
e). e) Hypotonia
FFTTT

21. Disorders of mitochondrial inheritance-
a). a) Barth syndrome
b). b) Leber hereditary optic
neuropathy
c). c) Cystic fibrosis
d). d) Myoclonic epilepsy
e). e) Multiple sclerosis
TTFTF

22. Examples of deletion disorders
a). a) Cri-du-chat syndrome
b). b) Angelman syndrome
c). c) Edward syndrome
d). d) Patau syndrome
e). e) Prader-willi syndrome
TTFFT

23. What are the invasive techniques of prenatal testing?--
a). a) Maternal serum screening
b). b) Combined test
c). c) Amniocentesis
d). d) Chorionic villous sampling
e). e) Ultrasonogram
FFTTF

24. Single gene disorders associated with enzyme defects are--
a). a) Phenylketonuria
b). b) Congenital adrenal hyperplasia
c). c) Sickle cell anemia
d). d) Galactosemia
e). e) Albinism
TTFTT

25. Clinical features found in Marfan syndrome-
a). a) Pectus excavatam
b). b) Hyperextended thumb
c). c) Increased upper segment and lower segment ratio
d). d) Increased elbow extension
e). e) Ectopia lentis
TTFFT

26. A patient came to see you complaining of arthritis. You find that he has a heart murmur and weak circulation to his extremities. You also notice that he is 6'7'' tall with extremely long fingers. You suspect an autosomal dominant disease and record a thorough family history from the patient. You think the cause of such condition is mutations in the gene which encode
a). a) Collagen
b). b) Ankyrin
c). c) Elastin
d). d) Spectrin
e). e) Fibrillin
EEEEE

27. Potential gene therapy methods for cancer include all of the following EXCEPT--
a). a) Inhibition of fusion proteins
b). b) Stimulation of the immune system
c). c) Increased expression of the angiogenic factors
d). d) RNA inheritance
e). e) Antisense oligonucleotides
CCCCC

28. Kevin is a 12yrs old boy with progressive muscle weakness. His motor skills were delayed since birth. Symptoms critical to his diagnosis were calf enlargement, toe walking and positive Gower sign. What condition does Kevin most likely have? --
a). a) Klinefelter syndrome
b). b) Cerebral palsy
c). c) Duchenne muscular dystrophy
d). d) Marfan syndrome
e). e) Osteogenesis imperfecta
CCCCC

29. Both husband and wife have normal vision though their fathers were color blind. The probability of their daughter becoming color blind is--
a). a) 0%
b). b) 25%
c). c) 50%
d). d) 75%
e). e) 100%
AAAAA

30. In human beings, multiple genes are involved in the inheritance of-
a). a) Color blindness
b). b) Congenital heart defects
c). c) Glucose-6-phosphate dehydrogenase deficiency
d). d) Nephrogenic diabetes insipidus
e). e) Polydactyly
BBBBB

31. A diseased man marries a normal woman. They get three daughters and five sons. All the daughters were diseased and sons were normal. The pattern of this disease is-
a). a) Autosomal dominant
b). b) Autosomal recessive
c). c) X-linked disorder
d). d) Cytogenic disorder involving autosome
e). e) Cytogenetic disorder involving sex chromosome
CCCCC

32. A 42yrs old male pt. presents with jerky, random and uncontrollable movements or chorea. Psychomotor functions have become increasingly impaired over the past few years. Common features are physical instability, abnormal facial expression and difficulties in chewing, swallowing and speaking. The patient has one parent who died at 47yrs after exhibiting similar symptoms. This case may be----
a). a) Phenylketonuria
b). b) Wilson disease
c). c) Huntington disease
d). d) Muscular dystrophy
e). e) Cystic fibrosis
CCCCC

33. A Cytogenetic disorders involving autosome is--
a). a) Turner syndrome
b). b) Kinefelter syndrome
c). c) own syndrome
d). d) Fragile X syndrome
e). e) Triple X syndrome
CCCCC

34. This condition is characterized by an accumulation of thick mucus in multiple organs including the lungs and pancreas. People with the condition often have chronic respiratory disease and digestive problems. What disease is it?--
a). a) Sickle cell anaemia
b). b) Coronary artery disease
c). c) Maple syrup urine disease
d). d) Cystic fibrosis
e). e) a1 antitrypsin deficiency
DDDDD

35. What condition results in abnormally shaped red blood cells which can lead to anaemia, fatigue, growth delay, jaundice, pain crises, frequent infections, pulmonary hypertension and organ damage?
a). a) Diabetes mellitus
b). b) Tay-sachs disaese
c). c) Hereditary spherocytosis
d). d) Sickle cell disease
e). e) Erythropoietic porphyria
DDDDD

36. A mutation in a codon leads to the substitution of one amino acid with another. What is the name for this type of mutation?-
a). a) Nonsense mutation
b). b) Missensense mutation
c). c) Frame shift mutation
d). d) Promoter mutation
e). e) Operator mutation
BBBBB

37. In which stage of meiosis in chromosomes pair and cross-over is-
a). a) Prophase I
b). b) Metaphase I
c). c) Prophase II
d). d) Metaphase II
e). e) Anaphase II
AAAAA

38. The polymerase chain reaction (PCR) is a technique that-
a). a) used to demonstrate DNA as the genetic material
b). b) is used to determine the content of minerals in a soil sample
c). c) uses short DNA primers and a thermostable DNA polymerase to replicate specific DNA sequences in vitro.
d). d) measures the ribosome transfer rate during translation
e). e) detects the level of polymerases involved in replication
CCCCC

39. Choose the correct statement about the genetic code --
a). a) Includes 61 codons for amino acids and 3 stop codons.
b). b) Almost universal, exactly the same in most genetic systems.
c). c) Three bases per codon.
d). d) Some amino acids are coded by multiple codons.
e). e) All of the above.
EEEEE

40. Which of the following disorders does not show X-linked inheritance?
a). a) Chronic granulomatous disease
b). b) Tay-sachs disaese
c). c) Alport syndrome
d). d) Hemophilia A
e). e) Hemophilia B
BBBBB

41. DNA ligase is-
a). a) an enzyme that joins fragments in normal DNA replication
b). b) An enzyme involved in translation
c). c) An enzyme that facilitates transcription of specific genes
d). d) An enzyme of bacterial origin which cuts DNA at defined base sequences
e). e) An enzyme which limits the level to which a particular nutrient reaches.
AAAAA

42. The occurrence of Down syndrome is mainly caused by--
a). a) Missense mutation in the SOD1 gene on chromosome 21
b). b) Mal-segregation at meiosis
c). c) Reciprocal translocation
d). d) Robertsonian translocation
e). e) Methylation aberration
BBBBB

43. Replication of DNA--
a). a) Takes place in a 'conservative' manner.
b). b) Takes place in a 'dispersive' manner.
c). c) Takes place in a 'semi-conservative' manner.
d). d) Usually involves one origin of replication per chromosome in eukaryotes.
e). e) Takes place only in the 3' to 5' direction.
CCCCC

44. Hemophilic man marries a normal woman. Their offspring will be--
a). a) All normal
b). b) All hemophilic
c). c) All girls- hemophilic
d). d) All boys- hemophilic
e). e) All offspring will die in infancy
AAAAA

45. A 15-year-old girl has developed multiple nodules on her skin over the past 10 years. On physical examination, there are 20 scattered, 0.3-cm to 1-cm, firm nodules on the patient's trunk and extremities. There are 12 light brown macules averaging 2 to 5 cm in diameter on the skin of the trunk. Slit-lamp examination shows pigmented nodules in the iris. A sibling and a parent are similarly affected) Genetic analysis shows a loss-of-function mutation. Which of the following inheritance patterns is most likely to be present in this family?
a). a) Autosomal dominant
b). b) Autosomal recessive
c). c) Mitochondrial
d). d) Multifactorial
e). e) X-linked recessive
AAAAA

46. A 38-year-old woman gives birth at 35 weeks' gestation to a female infant. Physical examination of the infant soon after delivery shows rocker-bottom feet, a small face and mouth, and low-set ears. On auscultation of the chest, a heart murmur is detected) The appearance of the infant's hands is shown in the figure) The infant dies at 4 months of age) Which of the following karyotypes was most likely present in this infant?
a). a) 45,X
b). b) 46,XX
c). c) 47,XX,+18
d). d) 47,XX,+21
e). e) 48,XXX
CCCCC

47. Which of the following abnormality of baby causes reduction of α-fetoprottein of maternal blood-
a). a) Trisomies
b). b) Wrong gestational age
c). c) Multiple pregnency
d). d) IUFD
e). e) Renal anomalies
AAAAA

48. A tall female with primary infertility presented to you with small breast with spare pubic hair on ex-genitalia & there is vagina with absent uterus and ovaries. What is the diagnosis:
a). a) Marfan syndrome
b). b) Gonadal dysgenesis
c). c) Testicular feminization syndarome
d). d) Turner syndrome
e). e) None is true
CCCCC

49. A newly described neurologic disorder is found to affect multiple family members in three generations that were available for study. In the first generation, two sisters and one brother were affected. In the second generation, all of the children of the first-generation sisters were affected, but none of the descendants of the first- generation son. In the third generation, all of the children of the affected second-generation women were affected, but none of the descendants of the secondgeneration men. The mode of inheritance exemplified here is
a). a) autosomal dominant
b). b) autosomal recessive
c). c) mitochondrial
d). d) X-linked dominant
e). e) X-linked recessive
CCCCC

50. The parents of a 17-year-old boy with Down syndrome seek counseling because they are concerned that their son may develop a life-threatening disorder known to be associated with his chromosomal abnormality. The physician should be prepared to iscuss which of the following disorders in terms of its association with Down syndrome?
a). a) Berry aneurysm of the circle of Willis
b). b) Creutzfeldt-Jakob disease
c). c) Lymphoblastic leukemia
d). d) Medullary carcinoma of the thyroid
e). e) Osteosarcoma
CCCCC