GENESIS

(Post Graduation Medical Orientation Centre)

Foundation-1 Batch

Total Number- 60 Pass Mark-42

b) Cystic fibrosis

d) Diabetes insipidus

a) Familial hypercholesterolemia

e) Duchenne muscular dystrophy

FTTFF (Robbin's 9th/Page-141-142)

c) Spinal muscular atrophy

Subject: Genetics

Question 16-30 is based on Single answers

6. Regarding X-Linked recessive trait

- a) Trait manifests only in male
- b) The gene responsible for this condition is transmitted from an affected man to all his daughters

Time: 20 Min

Date: 21/12/19

- c) The gene is never transmitted directly from father to son
- d) A carrier female will transmit the condition to 25% of her sons
- e) Normal offspring may be produced if mating occurs between an affected male and carrier female

FTTFT (Robbin's 9th/Page-142)

2. Diseases associated with Amyloidosis

1. Examples of autosomal recessive disorders are

- a) Chronic renal failure
- b) Thymic neoplasia
- c) Multiple myeloma
- d) Alzheimer disease
- e) Kaposi sarcoma

TFTTF (Ref. Robbin's 9th P-259 table:6.17)

3. DNA Amplification occurs

- a) PCR
- b) Recombinant DNA & Cloning
- c) DNA microassay
- d) Southern Blot
- e) FISH

TTFFF (Robbin's 9th/Page-175-176)

4. About Turner's syndromes

- a) It is the most common autosomal abnormality of female
- b) Mental status of the patient usually normal
- c) Amenrrhoea is one of most common feature
- d) Peripheral lymphoedema is seen at birth
- e) Mosaicism is not responsible for it

FTTTF

5. Mutation is

- a) Temporary change in DNA
- b) Production of mRNA
- c) Synthesis of new DNA
- d) Permanent change in DNA
- e) Protein malsynthesis

FFFTF (Robbin's 9th/Page-138)

7. Following are the X linked recessive diseases

- a) Myotonic dystrophy
- b) Color blindness
- c) Hemophilia
- d) Cranial diabetes insipidus
- e) Cystic fibrosis

FTTFF (Robbin's 9th/Page-141-142)

8. Diseases associated with polymorphism are

- a) Gout
- b) Colorectal carcinoma
- c) Breast carcinoma
- d) IBD
- e) IHD

TTTTF

9. Autosomal recessive disorders are

- a) Hunter syndrome
- b) Werdering Hoffman disease
- c) Colour blindness
- d) Adrenal hypoplasia
- e) Sickle cell anemia

FTFFT (Robbin's 9th/Page-141-142)

10. Regarding mutation

- a) Temporary change in DNA
- b) Gain or loss of chromosome set occurs in point mutation
- c) β-thalassaemia is a example of non sense mutation
- d) Visible change in chromosome occurs
- e) Fragile X syndrome affects coding regions

FFTFF (Robbin's 9th/Page-138)

11. Trinucleotide repeat disorders affecting coding regions are

- a) Myotonic dystrophy
- b) Kennedy disease
- c) Machado joseph disease
- d) Friedreich ataxia
- e) Progressive myoclonic epilepsy

FTTFF (Robbin's 9th/Page-168)

12. Feature/s of turner syndrome is/are

- a) A chromosome number of 45XY
- b) Secondary amenorrhoea
- c) Coarctation of aorta
- d) Multicystic ovaries
- e) Scanty pubic hair

FFTFT (Robbin's 9th/Page-166)

13. Characteristics of autosomal dominant disorders

- a) Two copies of the gene must be mutated for a person to be affected
- b) One mutated copy of the gene will be necessary for a person to be affected
- c) There is generation gap
- d) Defect in structural protein/receptor
- e) Late in onset

FTFTT (Robbin's 9th/Page-140)

14. Structural abnormalites of chromosome are

- a) Deletion
- b) Inversion
- c) Isochromosome
- d) Robertsonnian translocation
- e) Mosaicism

TTTTF(Robins, 9th / Page-160)

15. The DNA double helix is formed by the following base pairs-

- a) Adenine –thymine
- b) Adenine Uracil
- c) Cytosine –Guanine
- d) Glanine- Adenine
- e) Guanine Uracil

TETEE

Each question below contains five suggested answers- choose the <u>one best</u> response to each question (16-30)

16. Following are the autosomal dominant disorders except

- a) Huntington's disease
- b) Myotonic dystrophy
- c) Tuberous sclerosis
- d) MEN -1
- e) Albinism

E (Robbin's 9th/Page-141-142)

17. Most common cardiac defect in Turner's syndrome is

- a) Aortic stenosis
- b) Aortic regurgitation
- c) Coarctation of aorta
- d) Bicuspid aortic valve
- e) VSD

D (MR Khan 5th / Page-433)

18. Most common causes of Downs Syndrome

- a) Non-disjunction
- b) Mosaicism
- c) Robertsonian translocation
- d) Chromosomal deletion
- e) Chimerism

A (T-94%) (MR Khan 5th / Page-431)

- 19. A 25 yrs male has a work-up for infertility and found to have oligospermia. Physical examination findings include bilateral gynecomastia, reduced testicular size, and reduced body hair. Karyotypic analysis will most likely reveal which of the following abnormalities?
- a) 46,X,i(Xq)
- b) 47,XYY
- c) 47,XXY
- d) 46XX/47XX,+21
- e) 46XY, del(22q11)
- C (Robbin's 9th/Page-165)
- 20.A female patient presents with reduced stature, webbed neck, shield-shaped chest, renal malformation, diminished IQ and aortic coarctation. The parents informs you that they were diagnosed as a child with a chromosomal aneuploidy. You suspect:
- a) Down Syndrome (Trisomy 21)
- b) Edward Syndrome (Trisomy 18)
- c) Kleinefelter Syndrome (47, XXY)
- d) Turner Syndrome (45, X 0)
- e) Patau's syndrome
- D (Robbin's 9th/Page-166)

21. Which one of following is not characteristics of ADD?

- a) No new mutation
- b) Less severe than ARD
- c) Can exhibit anticipation
- d) Both male & female can transmit the disease
- e) Unaffected individual can transmit disease

A (Robbin's 9th/Page-140)

22. Autosomal Recessive Disorder are except

- a) Cystic fibrosis
- b) Phenylketonuria
- c) Wilson disease
- d) Friedreich's ataxia
- e) Lesch-Nyhan syndrome

F

23. Chromosome 9 is the site for

- a) Rh blood group
- b) Tuberous sclerosis
- c) Parathyroid hormone
- d) Red hair color
- e) Gulcagon

В

24. False about Barr body is?

- a) Genetically inactive
- b) Absent in male
- c) Situated centrally
- d) During cell division its disappear
- e)Inactive counterpart of two X chromosome

C (Khaleque /Page-90)

25. Which of following disease congenital heart disease doesn't occur

- a) Turner syndrome
- b) Klinefelter's syndrome
- c) Edward's syndrome
- d) Down syndrome
- e) Noonan syndrome

В

26. Which of following disease is a result of chromosomal number mutation

- a) Colorectal carcinoma
- b) Rettuoblastphoma
- c) Angelman syndrome
- d) Turner syndrome
- e) MEN

D

27. Feature related with autosomal Dominant disorder-

- a) More severe than Autosomal Recessive
- b) Enzyme dominancy is seen
- c) Variable expression is noted
- d) Occurrence of disease with a late onset in successive generation
- e) Skipping of generation is not seen

C (Robbin's 9th/Page-141-142)

28. Genetic code's character is not

- a) Unambigous
- b) Universality
- c) Non-Redundant
- d) Non-overlapping
- e) Unpunctuated

C

29. A patient suffers from hemophilia who marries a girl who is unaffected. On their probable children and generation which is not related on his marriage with his unaffected wife

- a) None of their daughter will be affected
- b) All son become healthy
- c) 100% of daughter become carrier
- d) All of his sister will be carrier
- e) His maternal grandfather could have had hemophilia

D

30. Disease where Gene therapy has not been attempted

- a) Sickle cell anaemia
- b) Cystic fibrosis
- c) Duchenne muscular Dystrophy
- d) Adenosine deaminase deficiency
- e) Hemophilia B

E