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PATHOLOGY EXAM # 3
December 10, 1999

Practical Questions:

1. This abdominal mass in a 3 year old boy is a:

- A. Ganglioneuroma
- B. Retinoblastoma
- C. Neuroblastoma
- D. Wilm's tumor - d 11p

2. This karyotype is associated with:

- A. Wilm's tumor
- B. Neuroblastoma | 76 minutes Abd, Adrenal d1p
- C. Cystic fibrosis ?
- D. Sudden Infant Death Syndrome

3. Pedigree showing the following mode of inheritance:

- A. dominant inheritance
 - B. recessive inheritance
 - C. X-linked dominant inheritance
 - D. mitochondrial DNA inheritance
- Mom → Kids*

4. Pedigree showing the following mode of inheritance:

- A. recessive inheritance
 - B. dominant inheritance
 - C. X-linked dominant inheritance
 - D. X-linked recessive inheritance
- every generation*
- B

5. This slide shows poor calcification of bone in a growing child due to a deficiency of vitamin D. What is the name of this deficiency disease?
- A. Osteoporosis
 - B. Rickets
 - C. Scurvy
 - D. Osteoarthritis

Written Questions:

6. Hirschprung's disease leads to:
- A. Functional obstruction and colonic dilatation distal to the affected segment
 - B. Functional obstruction and colonic dilatation proximal to the affected segment
 - C. Functional dilatation and colonic obstruction distal to the affected segment
 - D. Functional dilatation and colonic obstruction proximal to the affected segment
7. Factors associated with Sudden Infant Death Syndrome are:
- A. Low birth weight
 - B. Smoking and/or maternal drug abuse
 - C. Prematurity
 - D. Product of a multiple birth
 - E. All of the above
8. The Wilm's tumor is NOT associated with:
- A. WAGR syndrome
 - B. Denys-Drash syndrome
 - C. Beckwith-Wiedeman syndrome
 - D. n-myc oncogene amplification *Neuroblastoma*
9. The most serious complications from cystic fibrosis stem from its:
- A. Pancreatic involvement
 - B. Liver involvement
 - C. Pulmonary changes
 - D. Salivary gland changes

10. The most common karyotype found in tissue from a spontaneous first trimester miscarriage would be:

45X0

- B
- A. 47, XX, +21
 - B. 45, X
 - C. 45, X/ 46, XY
 - D. 46, XX

11. Individuals with the Turner syndrome 45,X

- A
- A. Will pass the condition to one half their children *sterile - streak gonads*
 - B. Are currently the patients most frequently on growth hormone treatment
 - C. Need to have their rudimentary testicular tissues removed early to prevent cancer
 - D. Are frequently encouraged to utilize their unusual facility for spacial relationships as architects

12. The correct nomenclature for a Down syndrome boy with a translocation between chromosomes 14 and 21 is:

- A. 46,XY,-14,t(14;21)
- B. 47,XY,+21
- C. 47,XY,-14,t(14;21)
- D. 45,XY,t(14;21)
- E. None of the above

13. A nineteen year old female with short stature, wide spaced nipples, and primary amenorrhea most likely has the karyotype of:

- A. 47,XX,+18
- B. 46,XY
- C. 47,XXY
- D. 45,X
- E. None of the above

14. Which of the following procedures is used as a routine technique for karyotyping using light microscopy?

- A. C-banding
- B. Fluorescence in situ hybridization (FISH)
- C. G-banding
- D. Q-banding
- E. BUdR-staining

15. Which of the following procedures is used for detection of specific DNA sequences in interphase nuclei or chromosomes?

- A. C-banding
- B. Fluorescence in situ hybridization (FISH)
- C. G-banding
- D. Q-banding
- E. BUdR-staining

16. Which of the following syndromes is associated with maternal disomy for chromosome 15?

- A. Hydatidiform mole
- B. Prader Willi syndrome
- C. Angelman syndrome - *Paternal*
- D. Klinefelter's syndrome
- E. Turner's syndrome

Both from Mom → PW
Both from Dad → Angelman

17. Which of the following syndromes is associated with the chromosome complement of 47,XXY?

- A. Hydatidiform mole
- B. Prader Willi syndrome
- C. Angelman syndrome
- D. Klinefelter's syndrome
- E. Turner's syndrome

18. The majority of cases of Down syndrome occur as a result of:
- A. Non-disjunction during maternal meiosis ✓
 - B. Mosaicism of normal and trisomic cell lines
 - C. Unbalanced 14:21 translocations
 - D. End to end fusion of two chromosomes 21
 - E. Non-disjunction during paternal meiosis
19. Chromosomes 1, 3, 19 and 20 are:
- A. Metacentric
 - B. Submetacentric
 - C. Acrocentric
 - D. Telocentric
 - E. None of the above
20. One would expect that routine cytogenetic analysis using standard G banding at normal levels of resolution could detect most cases of:
- A. Cru du Chat syndrome
 - B. Prader Willi syndrome
 - C. Angelman syndrome
 - D. Miller Deiker syndrome
 - E. DiGeorge syndrome
21. Which of the following statements is true?
- A. Conversion from a pre-mutation to a full-mutation for fragile X syndrome occurs in male meiosis
 - B. Female carriers for fragile X syndrome are easily diagnosed by chromosome analysis
 - C. The fragile X chromosome occurs in all cells of a male affected with the disorder but only in some cells of a transmitting male
 - D. Fragile X syndrome is the most common inherited cause of mental retardation

22. Which vitamin/mineral has been associated with a decreased risk of neural tube defects in the developing fetus:
- A. Iron
 - B. Folic acid
 - C. Riboflavin
 - D. Vitamin B₁₂
23. Individuals with Hurler disease (MPS I H):
- A. Usually are male
 - B. Have a single abnormal gene locus and a complimentary normal one
 - C. Are likely to show markedly reduced penetrance
 - D. Generally have two heterozygous parents
24. A relatively new genetic concept is that of "imprinting" of genes which means:
- A. The same gene may have a very different effect depending on whether it is inherited on a paternal or a maternal chromosome
 - B. Genes tend to accumulate more errors in each generation thus getting worse
 - C. Oncogenes are expressed only when moved to heterochromatic regions
 - D. Such conditions are most likely to occur in first born males
25. The following conditions would be associated with a 3-5% recurrence risk (chance of a second child with the same condition):
- A. Achondroplasia, cleft lip, color-blindness
 - B. Cystic fibrosis, sickle cell disease, pyloric stenosis
 - C. Anencephaly, cleft lip, pyloric stenosis
 - D. Spina bifida, achondroplasia, sickle cell disease
26. Dominantly inherited diseases, as compared to recessive disease, are most likely to:
- A. Be the result of new mutations
 - B. Result in single enzyme deficiencies
 - C. Have ethnic distributions
 - D. Decrease in frequency with increased paternal age

27. Nutritional assessment involves:
- A. Anthropometric measurements
 - B. Biochemical tests
 - C. Clinical observations
 - D. Data from diet history
 - E. All of the above
28. The term "primary" undernutrition indicates:
- A. Inadequate intake of one or more nutrients
 - B. Nutrient deficiency due to excess nutrient loss through blood or urine
 - C. Nutrient deficiency due to nutrient malabsorption
 - D. Nutrient deficiency due to increased metabolic needs
29. Secondary malnutrition may be the result of:
- A. A lack of available food
 - B. Missing nutrients in the food supply
 - C. Malabsorption
 - D. Limited food intake
30. A clinical sign of marasmus might include:
- A. Peripheral edema
 - B. Severe loss of muscle and fat stores
 - C. Hyperpigmented flaky dermatosis
 - D. Enlarged fatty liver
31. Edema and low serum protein levels are present in:
- A. Osteomalacia
 - B. Kwashiorkor
 - C. Obesity
 - D. Pellagra

32. Which of the following is NOT a clinical finding associated with kwashiorkor?
- A. Body mass index of >30
 - B. Enlarged fatty liver
 - C. Edema
 - D. Low serum proteins
33. Wernicke-Korsakoff syndrome is an encephalopathy-psychosis associated with:
- A. Deficiency of thiamine / excess alcohol
 - B. Excess iron
 - C. Excess vitamin A
 - D. Beri beri
34. An early clinical symptom of a vitamin C deficiency is:
- A. Impaired night vision
 - B. Hyperpigmented dermatosis
 - C. Bleeding gums
 - D. Hypersegmented granulocytes
35. A deficiency of thiamin targets which of the following tissues?
- A. Heart, peripheral nerves, and brain
 - B. Hair and skin
 - C. Skeletal system
 - D. All of the above
36. Skeletal changes can be seen in children with scurvy because of:
- A. Defective synthesis of osteoid matrix
 - B. Inadequate mineralization of cartilage
 - C. Epithelial metaplasia
 - D. All of the above

37. The following vitamin is a lipid soluble antioxidant that protects cell membranes from peroxidation:
- A. Thiamin
 - B. Niacin
 - C. Riboflavin
 - D. Vitamin E
38. A poor intake of vitamin K and reduced gut microflora may result in:
- A. A need for parenteral nutrition
 - B. Hypothrombinemia
 - C. Hyperkeratosis
 - D. All of the above
39. People with renal disease show decalcification of bone due to:
- A. Poor vitamin D intake
 - B. Lack of hydroxylation to an active form of vitamin D
 - C. Excessive urination
 - D. Poor intake of thiamin
40. The form of vitamin A active in vision is:
- A. Opsin
 - B. Tryptophan
 - C. Retinol
 - D. Keratin
 - E. Collagen
41. As far as is known, vitamin A does not play an important role in:
- A. Blood clotting
 - B. The synthesis of visual pigment
 - C. Maintaining mucous membrane
 - D. Maintaining the skin
 - E. The growth of bones and teeth

42. Vitamin E deficiency causes:
- A. Xerophthalmia A
 - B. Rickets D
 - C. Erythrocyte hemolysis
 - D. Osteomalacia D
43. The effects of a deficiency of vitamin D are most readily observed in the:
- A. Nervous system
 - B. Skeletal system
 - C. Muscular system
 - D. Epithelial system
44. The liver requires vitamin K in order to synthesize:
- A. Insulin
 - B. Calcitonin
 - C. Hemoglobin
 - D. Opsin
 - E. Prothrombin
45. Which symptoms would indicate a vitamin D deficiency?
- A. Slow blood clotting
 - B. Rupture of red blood cells
 - C. Bowed legs
 - D. Frequent respiratory infections
 - E. Abnormally high blood calcium level
46. Excessive intakes of vitamin D by adults may result in:
- A. A greater bone density
 - B. Mineral deposits in soft tissues such as the kidney
 - C. Deformity of leg bones, ribs, and skull
 - D. Increased bone calcification
 - E. A and D

47. An individual eating according to the U.S. Dietary Guidelines would consume his calories as:

- A. 55% Protein; 30% Fat; 15% Carbohydrate
- B. 15% Protein; 30% Fat; 55% Carbohydrate
- C. 55% Protein; 15% Fat; 30% Carbohydrate
- D. 30% Protein; 15% Fat; 55% Carbohydrate

48. A 50 year old man, 5'7" tall weighing 220 lbs. presents wanting to lose weight. He currently consumes 3000 kcals per day. In order to lose 2 lbs. per week, how many kcalories would you recommend?

- A. 1200 kcals
- B. 1500 kcals
- C. 1800 kcals
- D. 2000 kcals
- E. 2200 kcals

49. If the man in the above question has an ideal weight of 148 lbs. , how many grams of protein does he need each day?

- A. 54 grams
- B. 67 grams
- C. 80 grams
- D. 93 grams

$$\begin{array}{r}
 22 \overline{) 1480} \\
 \underline{132} \\
 160 \\
 \underline{154} \\
 60 \\
 \underline{60} \\
 0
 \end{array}$$

$$\begin{array}{r}
 67.3 \\
 \underline{178.4} \\
 67.3 \\
 \underline{178.4} \\
 0
 \end{array}$$

$$\begin{array}{r}
 67 \\
 \underline{18} \\
 53.84
 \end{array}$$

$$\begin{array}{r}
 2.2 \overline{) 1480} \\
 \underline{132} \\
 160 \\
 \underline{154} \\
 60 \\
 \underline{60} \\
 0
 \end{array}$$

50. According to the evidence-based federal guidelines for the treatment of obesity, what is the best approach?

- A. Reduce body weight by at least 2 BMI increments in 6 months
- B. Maintain newly reduced body weight for 6 months
- C. Encourage moderate physical exercise, 30 minutes or more on most days of the week
- D. All of the above