

6. CONGENITAL/ENDOCRINE

1. A 2-months old baby who has ambiguous genitalia presents to the ED with vomiting. Labs: Na⁺ = 125mmol/L, K⁺ 6mmol/L. What is the most likely diagnosis?

- a. Fragile X syndrome
- b. Turners syndrome
- c. Noonan syndrome
- d. Congenital adrenal hyperplasia
- e. Klinefelter's syndrome

2. An 8-months old infant presented with Failure to thrive and constipation. Exam: large tongue and history of prolonged neonatal jaundice. What is the most likely diagnosis?

- a. Down's syndrome
- b. Fragile X syndrome
- c. Prader willi syndrome
- d. DiGeorge syndrome
- e. Congenital hypothyroidism

3. A 6-year-old boy has been noticed to have problems with co-ordinating his voluntary movements over the last two years. He has a waddling gait and needs to support himself on his hands when rising from the floor. He has larger calves than other boys but he runs more slowly. Which is the most likely diagnosis?

- a. Myotonia
- b. Myasthenia gravis
- c. Duchenne muscular dystrophy
- d. Muscular atrophy
- e. Emery-Dreifuss muscular dystrophy

4. You are seeing a 6 year old female child for short stature although she is always hungry and eats a lot. There is history of delayed motor milestones but now mother is happy that she is like the rest of kids in family other than her weight gain because of increased oral intake. Which one of the following diagnoses can most likely explain her features?

a. Trisomy 21
 b. 45X
 c. Genetic deletion from long arm of chromosome
 d. 747XXY
 e. Deletion or disruption of genes in the proximal arm of chromosome 15

5. The parents of a 14-year-old boy are concerned about his short stature and lack of sexual development. By history, you learn that his birth weight and length were 3 kg and 50 cm, respectively, and that he had a normal growth pattern, although he was always shorter than children his age. The physical examination is normal. On plotting his height has been between 2nd and 10th centile all his life. His upper-to-lower segment ratio is 0.98. A small amount of fine axillary and pubic hair is present. There is no scrotal pigmentation; his testes measure 4.0 cm and his penis is 6 cm in length. In this situation, which of the following is the most appropriate course of action?

A. Measure pituitary gonadotropin
 b. Obtain a computed tomographic (CT) scan of the pituitary area
 c. Biopsy his testes
 d. Measure serum testosterone levels
 e. Reassure the parents that the boy is normal

6. A 5-month-old infant has been exclusively fed on normal infant formula. Upon introduction of fruit juices however, the child develops jaundice, hepatomegaly, vomiting, lethargy, irritability, and seizures. Tests for urine-reducing substances are positive. Which of the following is likely to explain this child's condition?

a. Tyrosinemia
 b. Galactosemia
 e. Hereditary fructose intolerance
 d. α1-Antitrypsin deficiency
 e. Glucose-6-phosphatase deficiency

7. Which of the following abnormalities is associated with short stature?

a. 45XO karyotype
 b. 47XXY karyotype
 e. 47XYY karyotype
 d. 46XY Karyotype
 e. Fragile X syndrome

8. A 17-years-old girl with short stature, neck webbing, and sexual infantilism is found to have coarctation of the aorta. A chromosomal analysis likely would demonstrate which of the following?

a. Mutation at chromosome 15q21.1
 b. Trisomy 21
 c. XO karyotype
 d. Defect at chromosome 4p16
 e. Normal chromosome analysis

9. Following effects of Vitamin D3 are false EXCEPT one:

a. Decreased phosphorus absorption
 b. Increase parathyroid hormone synthesis
 c. Increase parathyroid hormone secretion
 d. Decrease mineralization of the bones
 e. Increase calcium absorption

10. All of the following are the complications in the new born of a diabetic mother EXCEPT:

a. Hyperbilirubinemia
 b. Hyperglycemia
 c. Hypocalcemia
 d. Hypomagnesemia
 e. Hypoglycemia

11. You are seeing a 7 years old child in ward round. He is post head injury and with low GCS. His urine output is 5 ml/hour. His lab results are given below: Na-160, K-5, urea 80 (raised), glucose 80, creat 0.8 (normal), what do you think is wrong with him? Choose one correct answer?

a. Diabetes mellitus
 b. Diabetes Insipidus
 c. Urinary tract infection
 d. Fluid over load
 c. None of above

1.D	2.E	3.C	4.B	5.E	6.B
7.A	8.C	9.E	10.B	11.B	

12. Adrenogenital syndrome is associated with:

- a. sex-linked inheritance
- b. under-production of ACTH
- c. vomiting in the neonatal period
- d. undescended testicles
- e. convulsions in the neonatal period

13. Short stature is associated with:

- a. chronic renal disease
- b. malabsorption
- c. down's syndrome
- d. emotional deprivation
- e. all of the above

14. The following conditions are generally inherited by sex-linked recessive genes:

- a. duchenne muscular dystrophy,
- b. temporal lobe epilepsy.
- c. meningocele
- d. cystic fibrosis,
- e. all of the above

15. A boy with Down's syndrome is likely to grow up into a man who:

- a. has children half of whom will have Down's syndrome
- b. has an intelligence quotient of less than 50
- c. has a life expectancy less than a man with cystic fibrosis
- d. is above average height
- e. has recurrent seizures

16. A girl of two years presents with a two week history of apparent thirst. Her mother complained that she had become very irritable and feels the child is passing excess urine. Following admission to hospital and on a free fluid intake. A 24 hour urine collection amounts to 2360 ml. No abnormal constituents are found on urinalysis and culture is sterile. Which of the following is the most appropriate statement?

a. Congenital nephrogenic diabetes insipidus is the most likely diagnosis

b. Urine osmolality in excess of 285 mosmol/kg is to be expected

c. Computerized axial tomography of the brain is indicated

d. An overnight concentration test should be performed

e. The treatment regimen should include free access to fluids

17. Familial hypercholesterolaemia (hyperlipidaemia type 11:a)

- a. has an autosomal dominant transmission
- b. is caused by a failure of enzymatic breakdown of cholesterol
- c. is an uncommon example of inherited metabolic disease
- d. needs only to be treated in the homozygous state
- e. is asymptomatic in heterozygotes

18. Hyperthyroidism is characterized by:

- a. Bradycardia
- b. Intolerance to cold
- c. Generalized oedema
- d. Warm and sweaty skin
- e. Weight gain

19. One of the following is NOT a characteristic feature of Down's syndrome

- a. Brush field spots
- b. Clinodactyly
- c. Polydactyly
- d. Large gap between the first and second toes
- e. Simian crease

20. Metabolic acidosis is not a feature of the following

- a. Diarrhea
- b. Diabetic ketoacidosis
- c. Shock
- d. Starvation
- e. Vomiting

21. The most common cause of fits in Neonate of a Diabetic Mother is

- Hypomagnesemia
- Hyponatremia
- Hypokalemia
- Hypoglycemia
- Hypertension

22. Thalassemia is transmitted as:

- Autosomal dominant
- Autosomal recessive
- Multifactorial
- X-linked dominant
- X-linked recessive

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25. A 10 y/o boy is clinically obese and the shortest in his class. He had a renal transplant last year and his mother is worried that he is being bullied. What is the most probable diagnosis?

- Cushing's syndrome
- Congenital hypothyroidism
- Pseudocushing's syndrome

- Lawrence moon biedel syndrome
- Down's syndrome

26. A 6-year-old girl started wetting herself up to 6 times per day. What is the most appropriate treatment

- Sleep alarms
- Desmopressin
- Reassure
- Behavior training
- Imipramine.

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28. Being severely malnourished, a child is found to have bleeding gums and easy bruising along with numerous poorly healing skin ulcerations. Assuming that these findings resulted from vitamin C deficiency. What is the likely mechanism of these findings?

- Defective DNA synthesis
- Defective production of γ -aminobutyric acid
- Impaired carboxylation of coagulation factors II, VII, IX, and X
- Impaired hydroxyproline and hydroxylysine production
- Impaired renal la-hydroxylase

12.C	13.E	14.A	15.B	16.C
17.A	18.D	19.C	20.E	21.D
23.D	24.E	25.A	26.B	27.C

29. An 8 weeks old baby boy is noted to be jaundiced. He is breast-feeding well and has gained 300g since birth. His stools are yellow and his urine is pale straw colored. What is the most likely diagnosis?

- a. Galactosemia
- b. Biliary atresia
- c. G6PD deficiency
- d. Breast milk jaundice
- e. Congenital viral infection

30. Infant of diabetic mother with weight 3.8 kg presented with seizures after 16 hours of birth. What is the cause?

- a. Hypoglycemia
- b. Hypocalcemia
- c. Birth asphyxia
- d. Intraventricular hemorrhage
- e. All of the above

31. Kostmann's syndrome treatment is

- a. Anti-thymocyte globulin + cyclosporine
- b. Anti-thymocyte globulin + cyclosporine + GM-CSF
- c. G-CSF
- d. GM-CSF
- e. Cyclosporin

32. Fetal alcohol syndrome is characterized by all except

- a. Microcephaly
- b. Low intelligence
- c. Normal behavior
- d. Septal defects of heart
- e. Poor memory

29.A

30.A

31.C

32.C