Exam 1 Questions

Q 1. The following are true of X-linked recessive disorders

A. Females are unaffected
B. Sons of affected males have a 100% chance of being affected
C. Sons of female carriers have a 25% chance of being affected
D. Daughters of affected males have a 100% chance of being carriers
E. Daughters of female carriers have a 50% chance of being carriers

Q 2. Recognized features of Marfan syndrome include

A. Lens subluxation, usually downwards
B. Femoral hernia
C. Learning disability
D. Hemivertebrae
E. Autosomal recessive inheritance

Q 3. Apert syndrome is characterized by

A. Syndactyly
B. Polydactyly
C. Acne
D. Irregular craniosynostosis
E. Mental deficiency

Q 4. In achondroplasia, the following problems may develop

A. Upper airway obstruction
B. Cord compression
C. Hydrocephalus
D. Osteoarthritis
E. Sensorineural deafness

Q 5. The following are true

A. Somatic cells contain 23 pairs of autosomes
B. Gametes are haploid
C. The bases cytosine and adenine pair together
D. Adenine is a pyrimidine nitrogenous base
E. In DNA replication, new base pairs are added to the 5′ end of the single DNA strand

Q 6. Regarding the optic (second) cranial nerve

A. It is derived embryologically from the hind-brain
B. It has both an intra-orbital and intracranial component
C. The fibres from the lateral half of the retina cross over at the optic chiasma to the optic tract of the opposite side
D. The majority of the fibres in the optic tract end in the medial geniculate body
E. The upper half of the retina is represented on the lower lip of the calcerian fissure of the visual cortex

Q 7. Parasympathetic nervous system stimulation causes

A. Stimulation of the detrusor muscle of the bladder
B. Pilo-erection
C. Uterine contraction
D. Bronchial constriction
E. Activation of peristalsis

Q 8. Regarding neural tube defects

A. There is an association with isotretinoin taken during pregnancy
B. The recurrence risk after two previous infants with neural tube defects is 10%
C. Reduced alpha-fetoprotein (α-FP) is present in the amniotic fluid
D. Spina bifida occulta is not associated with neurological symptoms
E. Neural tube closure takes place in the fourth week of intrauterine life

Q 9. Lennox-Gastaut syndrome

A. Presents with malabsorption
B. Is a cause of developmental regression

C. Responds well to vitamin supplementation
D. Has autosomal recessive inheritance
E. Is more common in Down's syndrome

Q 10. The following are side-effects of phenytoin

A. Hyperphagia
B. Peripheral retinal atrophy
C. Hirsutism
D. Acne
E. Rickets

Q 11. Concerning juvenile myoclonic epilepsy

A. The seizures are partial
B. A quarter of patients have a positive family history of epilepsy
C. The gene has been identified on chromosome 7
D. Phenytoin is the treatment of choice
E. Photic stimulation increases positive EEG findings by over 30%

Q 12. A normal 5-year-old is able to

A. Do all their buttons up
B. Hop
C. Tie shoelaces
D. Say their home address
E. Copy a cross

Q 13. Iris coloboma is seen in

A. Down's syndrome
B. Turner syndrome
C. Rubinstein-Taybi syndrome
D. Trisomy 13
E. Klinefelter's syndrome

Q 14. The following are causes of a large pupil

A. Holmes-Adie pupil
B. Congenital rubella syndrome
C. Lowe's oculocerebrorenal syndrome
D. Ecstacy
E. Fabry's disease

Q 15. The following are true of the oxyhaemoglobin dissociation curve

A. At the p50 the surrounding partial pressure of oxygen is normally 27 mmHg
B. In mixed venous blood the oxygen saturation is 50%
C. Methaemoglobinaemia decreases the affinity of haemoglobin for oxygen
D. Cyanotic congenital heart disease does not affect the curve
E. Hypothermia increases the affinity of haemoglobin for oxygen

Q 16. The following may be seen in the blood film in iron deficiency anaemia

A. Target cells
B. Anisocytosis
C. Pencil cells
D. Thrombocytopenia
E. Increased free erythrocyte porphyrin

Q 17. The following cause splenomegaly with resultant anaemia

A. Niemann-Pick disease
B. Langerhans cell histiocytosis
C. Portal hypertension
D. Thalassaemia major
E. Immune haemolytic anaemia

Q 18. These hereditary disorders predispose a child to thrombosis

A. Protein C deficiency
B. Hermansky-Pudlak syndrome
C. Antithrombin III deficiency
D. Bernard-Soulier syndrome
E. Factor V Leiden deficiency

Q 19. Concerning laboratory investigations of a bleeding disorder

A. Von Willebrand's disease has a prolonged APTT
B. Prothrombin time (PT) is prolonged in haemophilia B
C. Bleeding time is prolonged in haemophilia A
D. Prolonged bleeding time occurs with impaired platelet numbers

E. APTT is sensitive to factors in the intrinsic coagulation pathway

Q 20. The following conditions are primary T-cell deficiencies

A. Adenosine deaminase deficiency
B. Hereditary angioneurotic oedema
C. Wiskott-Aldrich syndrome
D. MHC class II deficiency
E. Chronic granulomatous disease

Q 21. In staphylococcal scalded skin syndrome

A. The child is generally well
B. It is usually due to *Staphylococcus aureus* group I phages
C. The skin loss is superficial
D. It is most common in school age children
E. Frozen section of skin may help the diagnosis

Q 22. Regarding galactosaemia

A. It causes an inability to metabolize galactose
B. It causes an inability to metabolize lactose
C. It is X-linked recessive
D. It causes dysarthria
E. Ovarian failure is a feature

Q 23. In Hartnup disease

A. It is usually asymptomatic in children
B. It is a cause of ataxia
C. There is a photosensitive rash
D. The acylcarnitine profile of a blood spot is diagnostic
E. There is an accumulation of tryptophan

Q 24. Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

A. Is a urea cycle defect
B. Affects up to 1 in 40 of the UK population as asymptomatic carriers
C. May present with encephalopathy
D. Causes hepatomegaly
E. Shows a characteristic urine amino acid profile

Q 25. In paediatric liver disease, portal hypertension

A. Occurs when portal pressure is elevated to 10–12 mmHg
B. Results in cephalic flow of collaterals inferior to the umbilicus
C. May cause signs of spinal compression
D. May result from factor V Leiden deficiency
E. Should be treated prophylactically with propranolol in children under the age of 4 years

Q 26. Hirschsprung's disease

A. Is inherited in an autosomal recessive fashion
B. Is due to unopposed parasympathetic activity in the affected segment of the bowel
C. Has an equal incidence in girls and boys
D. Is seen more frequently in children with Down's syndrome than in the general population
E. Presents in more than 80% in the neonatal period

Q 27. The following occur more commonly in Crohn's disease than ulcerative colitis

A. Erythema nodosum
B. Pyoderma gangrenosum
C. Ankylosing spondyloarthropathy with HLA B27
D. Uveitis
E. Cholangiocarcinoma

Q 28. Recognized features of Alagille's syndrome include

A. Tuberous xanthomas and raised serum cholesterol
B. Progression to cirrhosis and chronic liver failure requiring liver transplantation
C. Aortic stenosis
D. Tetralogy of Fallot
E. Abnormalities of peroxisomal function

Q 29. In chronic liver disease in childhood, the following are correct

A. Spironolactone is useful in the treatment of ascites at any age
B. A low plasma cholesterol is an adverse prognostic feature
C. Sleep reversal occurs as a feature of hepatic encephalopathy

D. There is an increased overall incidence of Hirschsprungs's disease
E. Spontaneous bacterial peritonitis is a potentially fatal complication of ascites

Q 30. In the fetal circulation

A. Oxygenated blood is transported to the fetus via the umbilical artery
B. The ductus venosus drains into the superior vena cava
C. Oxygenated blood passes through the foramen ovale from the right to the left side of the heart
D. There is no significant blood flow through the coronary arteries
E. Blood flows from the pulmonary artery via the ductus arteriosus into the descending aorta

Q 31. Chromosome 22q11 microdeletion is associated with the following forms of congenital heart disease

A. Truncus arteriosus
B. Patent ductus arteriosus
C. Dissecting aortic aneurysm
D. Peripheral pulmonary stenosis
E. Tetralogy of Fallot

Q 32. Torsades de Pointes may be caused by

A. Organophosphate poisoning
B. Aspirin
C. Anorexia nervosa
D. Hypomagnesaemia
E. Lead poisoning

Q 33. The following features would be consistent with the murmur of a ventricular septal defect

A. Wide fixed splitting of the second heart sound
B. Loudest at the upper left sternal edge
C. Mid-diastolic apical murmur
D. Parasternal thrill
E. Loud P2

Q 34. Congenital complete heart block

A. Always requires treatment with a pacemaker
B. Is associated with Turner syndrome
C. May be secondary to an atrioventricular septal defect
D. Is associated with maternal anti-Ro antibodies
E. May present with hydrops fetalis

Q 35. Causes of an acidosis with a normal anion gap are

A. Lactic acidosis
B. Ketoacidosis
C. Salicylate poisoning
D. Proximal renal tubular acidosis
E. Diarrhoea

Q 36. Causes of Fanconi's syndrome include

A. Hypoparathyroidism
B. Cystinosis
C. Glue-sniffing
D. Galactosaemia
E. Wilson's disease

Q 37. Haemolytic uraemic syndrome

A. Causes hypokalaemia
B. Involves a sideroblastic anaemia
C. May be caused by salmonella
D. If familial, has a better prognosis
E. Is the most common cause of renal failure in children in the UK

Q 38. The features of Alport syndrome are

A. Haematuria
B. Proptosis
C. Cataract
D. Asymmetrical craniosynostosis
E. Hyperphagia

Q 39. Regarding surfactant

A. It decreases lung compliance at low lung volumes
B. There is a lower incidence of pneumothorax with natural surfactant than synthetic surfactant

C. It decreases the need for oxygen in infants with respiratory distress syndrome
D. It can cause a transient decrease in oxygen saturation when given
E. It helps maintain the functional residual capacity of the lungs

Q 40. Cystic fibrosis

A. Causes a hyperkalaemic alkalosis
B. Is a cause of intussusception
C. Is due to a mutation in the cystic fibrosis transmembrane regulator (*CFTR*) gene on chromosome 8
D. Is a cause of nasal polyps
E. Is due to a defect in the sodium channels

Q 41. Oligoarticular juvenile idiopathic arthritis

A. Has an equal sex ratio
B. Goes on to affect more than four joints after 6 months in 50% of cases
C. Is rheumatoid factor (RhF) positive
D. Typically affects the hands and feet
E. If associated with uveitis, may progress silently to blindness

Q 42. Inguinal hernias

A. Are usually direct in children
B. Are more common in females in childhood
C. Are associated with Marfan syndrome
D. Are more common on the right side
E. Are associated with prematurity

Q 43. Octreotide

A. Is a GNRH analogue
B. Is used in the treatment of insulinoma
C. Causes irreversible alopecia
D. Commonly causes altered liver function tests
E. Is effective as an anti-emetic in palliative care

Q 44. Mycophenolate mofetil

A. Is a calcineurin inhibitor
B. Can cause hypertension

C. Has a lower incidence of side-effects in children
D. Is an inactive precursor
E. May cause pancreatitis

Q 45. The side-effects of doxorubicin include

A. Mucositis (uncommonly)
B. Supraventricular tachycardia
C. Cardiomyopathy at low dosage
D. Pulmonary fibrosis
E. Retinopathy

Q 46. Physical features seen in anorexia nervosa include

A. Acidosis
B. Low growth hormone
C. Low rT3
D. Raised LH
E. Short QT interval on ECG

Q 47. Regarding multiple pregnancy

A. The incidence of monozygotic twins is 1 in 1000 pregnancies
B. There is a 1 in 33 spontaneous rate for dizygotic twins
C. There is an increased risk of congenital anomalies among monozygotic twins
D. Dizygotic twins always have a dichorionic placenta
E. Dichorionic twins are at risk of twin-twin transfusion syndrome

Q 48. Neonatal pulmonary haemorrhage is associated with

A. Pneumonia
B. Acute cardiac failure
C. Respiratory distress syndrome
D. Prematurity
E. Asphyxia

Q 49. In osteopenia of prematurity

A. Inadequate milk intake is a risk factor
B. Large for gestational age is a risk factor
C. Clinical rickets is commonly seen