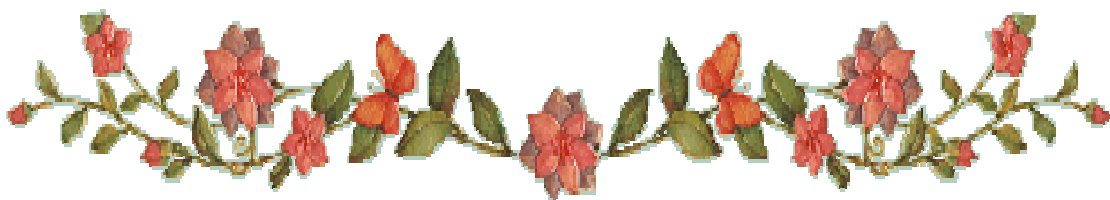


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Pediatric
Questions & Answers
Group 1 - 2008





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Question 1

An ostium premium ASD

- A- Results in right axis deviation(RAD) on the ECG.
- B- Is seen in Holt-Oram syndrome.
- C- Is the most common form of ASD.
- D- May be left to close spontaneously.
- E- Reverse splitting of the 2nd heart sound occurs.

Answer Question 1

A- False B- False C- False D- False E- False

An ostium premium results from failure of development of the septum premium. And there is usually also a cleft in the anterior leaflet of the mitral valve. Ostium secundum defects are the most common type of ASD, and there are seen in Holt-Oram syndrome. Ostium premium defects are associated with Down syndrome and Ellia-van-Creveld syndrome. The defect may be asymptomatic, but can result in heart failure and recurrent pneumonia. There is a pan-systolic mitral regurgitation murmur, and there is fixed wide splitting of the 2nd heart sound. These defects, unlike many VSDs, require surgical repair..

Question 2

In Wolf-Parkinson-White (WPW) syndrome

- A- There is a risk of ventricular tachycardia.
- B- Presentation may be with hydrops foetalis.
- C- Type a is seen in association with Ebstein's anomaly.
- D- Flecanide reduces the recurrence risk of tachycardias.
- E- The ECG shows a narrow QRS complex.

Answer Question 2

A- True B- True C- False D- True E- False

Wolf-Parkinson-White(WPW) syndrome is a congenital condition resulting from an abnormal connection between the atria and ventricles.

The ECG has a short PR interval. Wide QRS complexes and a delta wave. There are two types: (a) and (b).

Type (a) is the commonest, and is activation of the left ventricle via the accessory pathway. **Type (b)** activation of the right ventricle via the accessory pathway and occurs with Ebstein's anomaly.

Intermittent episodes of Supraventricular tachycardia occur, and there is a risk of VT.

Presentation may be with hydrops foetalis or intra-uterine death if there is in-utero tachycardia.

Flecanide can be used to stop the tachycardia and to reduce the risk of recurrence of tachycardia.





Question 3

In Tetralogy of Fallot

- A- Cyanosis is reduced by taking a hot bath.
- B- Cyanosis is often present in the first few days of life.
- C- There is a right sided aortic arch in about 50% of cases.
- D- The murmur becomes softer during a cyanotic spell as flow through the pulmonary valve is increased.
- E- There is an association with DiGeorge syndrome.

Answer Question 3

A- False B- False C- False D- False E- True

Tetralogy of Fallot is the most common congenital cyanotic heart condition. It may present with cyanosis in the first few days of life, though this is rare. Usually it presents as a murmur detected in first few months of life or as hypercyanotic spells in late infancy.

A hot bath will increase cyanosis, as the vasodilatation results in a decrease in the peripheral vascular resistance, and so the flow through the pulmonary artery decreases, and cyanosis increases. During a cyanotic spell the decrease (not increase) in flow through the pulmonary artery results in the murmur becoming softer or inaudible.

There is a right sided aortic arch in about 30% of cases.

Tetralogy of Fallot associated with DiGeorge syndrome , and with Down's syndrome, CHARGE syndrome and VACTERL syndrome.

Question 4

Regarding infective endocarditis

- A- It is commonly seen with atrial septal defects(ASD).
- B- It can be associated with aneurysms of the cerebral arteries.
- C- Osler's nodes are commonly seen.
- D- Splitter haemorrhages are present in about 25% of cases.
- E- It is caused by staphylococcus aureus in about half of the cases of acute endocarditis.

Answer Question 4

A- False B- True C- False D- False E- True

Infective endocarditis is seen particularly with congenital heart disease and on previously damaged and prosthetic valves.

It is very uncommon in atrial septal defects, and is seen with high flow defects.

Mycotic aneurysms can occur in both central and peripheral arteries.

Osler's nodes and splinter haemorrhages are both seen uncommonly.

Staphylococcus aureus causes about half of the acute cases. Streptococcus viridans causes about half of the cases of subacute bacterial endocarditis.





Question 5

Lymphocytic interstitial pneumonitis(LIP)

- A- Is uncommon in vertically acquired HIV infection.
- B- Is due to a protozoal infection.
- C- Is seen on chest X-ray as a perihilar infiltration in a butterfly distribution.
- D- May be associated with hepatosplenomegaly.
- E- Steroid therapy is always necessary.

Answer Question 5

A- False B- False C- False D- True E- False

Lymphocytic interstitial pneumonia (LIP) is a disease of unknown aetiology. It is seen most commonly in children who have vertically HIV infection. The chest X-ray findings are of a diffuse infiltration and sometimes a perihilar lymphadenopathy also. There may be associated hepatomegaly, generalised lymphadenopathy or pancreatitis. Steroid therapy is only necessary if symptomatic to reduce oxygen requirement and increase exercise tolerance.

Question 6

In cystic fibrosis

- A- The underlying defect in sodium channel function
- B- The immune reactive trypsin (IRT) may be checked on the Guthrie card.
- C- In most cases the genetic defect is a loss of Valine at position 508 on chromosome 7
- D- A sweat test with a sodium content above 40 mmol/l is diagnostic
- E- A false positive sweat test may be seen in Ectodermal dysplasia

Answer Question 6

A- False B- True C- False D- False E- True

In cystic fibrosis, the underlying defect is in a chloride channel blocker, cystic fibrosis transmembrane regular (CFTR). The chromosome defect is loss of phenylalanine at position 508 on chromosome 7 in around 80% of cases. The immune reactive trypsin (IRT), used in diagnosis of those under 3 months can be checked on the Guthrie card.

The sweat sodium concentration is the definitive test and this is diagnostic if on two tests the sodium is above 60mmol/l. False positive sweat tests are seen in Ectodermal dysplasia.





Question 7

Regarding severe respiratory compromise

- A- Subcostal and intercostal recession are more significant signs of respiratory compromise in infants than in school age children
- B- Respiratory failure is second only to cardiac failure as the commonest cause of cardiac arrest in children
- C- The loudness of the respiratory wheeze corresponds directly with the severity of the bronchospasm
- D- In inspiratory failure treatment should only be given after examination
- E- Oxygen given via nasal canula results in a constant inspired oxygen concentration.

Answer Question 7

A- False B- False C- False D- T False E- False

Infants have softer, more compliant ribs, resulting in recession more easily, therefore recession in an older child is more significant.

About 85% of cardiac arrests in children are due to respiratory failure.

The loudness of a wheeze is dependant on both airway diameter and flow. A softening of wheeze may indicate a tiring child (with reduced airway flow).

Basic resuscitation in inspiratory failure should be done prior to detailed examination.

Oxygen delivery is variable when given via nasal canula.

Question 8

Associations of Coeliac disease include

- A- Epilepsy with posterior cerebellar calcification
- B- IgA nephropathy
- C- Oesophageal carcinoma
- D- Addison's disease
- E- Sjogren's syndrome

Answer Question 8

A- True B- True C- True D- False E- True

IDDM and thyroiditis are endocrine associations but Addison's disease is not recognised as such, although there is a predominance of autoimmune diseases.





Question 9

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- It seen more frequently in children with Down's syndrome than in the general population
- E- Presents in more than 80% in the neonatal period

Answer Question 9

A- False B- False C- False D- True E- True

Polygenic inheritance, male>female. Increase in sympathetic activity is due to absence of parasympathetic ganglions in the plexi of Auerbach and Meissner.

Question 10

The following more commonly occur in Crohn's disease than Ulcerative colitis

- A- Erythema nodosum
- B- Pyoderma gangrenosum
- C- Ankylosing spondyloarthritis with HLA B27
- D- Uveitis
- E- Cholangiocarcinoma

Answer Question 10

A- True B- False C- False D- False E- True

Eye complications occur equally, sclerosing cholangitis, chronic active hepatitis, cirrhosis, and pericholangitis are all commoner in UC.

Question 11

Anorexia nervosa

- A- Is more common than bulimia
- B- Can have a low T4 in the presence of a normal T3
- C- Can demonstrate a raised growth hormone
- D- May have a hypochloreaemic hypokalaemic alkalosis
- E- Is associated with diarrhea more commonly than constipation

Answer Question 11

A- False B- False C- True D- True E- False

T3 is more often low with an apparently T4. Diarrhea may occur with laxative abuse but constipation is commonest with inherent hypomotility of the gut.





Question 12

The following are causes of haematuria in childhood

- A- Alport's syndrome
- B- E.coli 0157
- C- Factor V leiden deficiency
- D- Alpha thalassaemia
- E- Rapid increase in intracranial pressure.

Answer Question 12

A- True B- True C- True D- False E- False

Haemolytic uraemia syndrome may be triggered by E.coli 0157. Factor V leiden deficiency is present in up to 10% of the Caucasian population and results in a pro-coagulation tendency, which in turn may lead to renal thrombosis and haematuria.

Question 13

Henoch-Schönlein purpura

- A- Has a preponderance for late Winter and early Summer
- B- Exhibits a male-female ratio of 2 – 1
- C- Is associated with a raised IgA in over 40%
- D- May cause death from pulmonary haemorrhage
- E- Is associated with a prolonged bleeding time

Answer Question 13

A- True B- True C- True D- True E- False

IgA is raised in over 50%. It is a vasculitic phenomenon and this is the cause of the rash, rather than any abnormality of coagulation or platelet function which may lead to a prolonged clotting or bleeding time. Death from massive pulmonary haemorrhage is reported although rare. Opinion on the use of steroids in children with abdominal pain is still divided, as it was felt that they mask the pain associated with intussusception.

Question 14

Fanconi's syndrome

- A- Is associated with cystinuria
- B- Is usually associated with a haemoglobin <10g/dl
- C- Results in dehydration, polyuria and polydipsia
- D- Is acquired by glue-sniffing
- E- Can produce a urine with a pH of 5.0 and a metabolic acidosis.

Answer Question 14

A- False B- False C- True D- True E- True

Cystinosis is associated with Fanconi's syndrome. Do not get Fanconi's anaemia confused with Fanconi's syndrome.





Question 15

In a child with minimal change nephritic syndrome

- A- Penicillamine is a recognised cause
- B- A history in the previous 6 weeks of a streptococcal illness is likely
- C- The albumin : creatinine ratio is $> 200\text{mg}/\text{mmol}$
- D- Initial management is prednisone $90\text{mg}/\text{m}^2/\text{day}$
- E- Abdominal pain requires parenteral penicillin and a watch and wait policy

Answer Question 15

A- False B- False C- True D- False E- False

Penicillamine causes membranous GN. Most cases of minimal changes GN are idiopathic. Initial treatment is prednisone $60\text{mg}/\text{m}^2/\text{day}$ until on Proteinuria then alternate days for 4 weeks, but if no response then a renal biopsy is advocated. Abdominal pain is a medical emergency and may indicate hypovolaemia requiring urgent correction, or major vein thrombosis, or primary or pneumococcal peritonitis.

Question 16

In Alagill's syndrome the features which are recognised include

- A- Tuberous xanthomas and raised serum cholesterol
- B- Progression to cirrhosis and chronic liver failure requiring liver transplant
- C- Tubulointerstitial nephropathy
- D- Tetralogy of Fallot
- E- Abnormalities of peroxisomal function

Answer Question 16

A- True B- False C- True D- True E- False

If children with Alagille's syndrome, usually inherited as an Autosomal dominant condition, and also called arteriohepatic dysplasia, require liver transplantation it is due to uncontrollable itching and poor quality of life rather than liver failure which is not common, Zellweger's syndrome (cerebrohepatorenal syndrome) is an example of a peroxisomal disorder but this group does not include Alagille's syndrome. Features of Alagille's syndrome include progressive intrahepatic bile duct paucity; intense pruritis, typical facies with Hypertelorism, deep set eyes, long nose, broad forehead and small mandible, posterior embryotoxon of the eyes, peripheral pulmonary stenosis and Fallot's tetralogy; Tubulointerstitial nephropathy; butterfly vertebrae, tuberous xanthomas and raised serum cholesterol; and it occurs at an incidence of 1 in 100,000 live births. Gene mapping is now possible with a gene defect localised to chromosome 20p (gene termed JAG1) coding for a ligand of Notch 1, which is one of a member of 4 transmembrane proteins.





Question 17

In paediatric liver disease portal hypertension

- A- Occurs when portal pressure is elevated to 10-12mmHg
- 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 under the age of 12 years

Answer Question 17

A- True B- False C- True D- True E- False

Normal portal pressure is only 7 mmHg. Caput medusae results in blood flow away from the umbilicus. Perivertebral and perispinal collaterals may occur and in extremis may cause signs of spinal compression. Factor V leiden deficiency occurs in up to 10-15% of the Caucasian population and may result in hypercoagulation and hepatic or portal vein thrombosis. As younger children rely, in part, on an increase in their heart rate to counter hypovolaemia secondary to potential haemorrhage from varices, beta-blockers are not recommended to decrease portal pressure as this protective mechanism may therefore be compromised.

Question 18

In acute liver failure

- A- Aminotransferase levels are not predictive of outcome
- B- Due to sodium valporate therapy the chance for a neurologically intact outcome is very poor
- C- Hyperventilation usually accompanies stage II–III hepatic encephalopathy and may result in respiratory alkalosis
- D- Fluid restriction to <75% of maintenance is the key strategy in prevention of intracerebral oedema
- E- Coagulation support should only be used if active bleeding occurs or to cover invasive procedures

Answer Question 18

A- True B- True C- True D- True E- True

A drop in AST/ALT (SGOT/SGPT) can reflect massive hepatocellular necrosis and little residual viable liver tissue. A mitochondrial cytopathy uncovered by sodium valporate is usually responsible for Fulminant liver failure in this instance and therefore multisystem involvement including neurological precludes survival even with liver transplant in nearly all cases.

Metabolic acidosis may occur, but respiratory alkalosis also occur in stage II – III encephalopathy. Coagulation is a good indicator of liver function and should not be artificially supported unless necessary. Vitamin K and the liver's response to it may yield useful information.





Question 19

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 Hirschsprung's disease
- E- Spontaneous bacterial pneumonia is a potentially fatal complication of ascites.

Answer Question 19

A- True B- True C- True D- False E- True

Plasma cholesterol reflects liver synthetic function, like serum albumin and coagulation state, and if it is low this reflects worsening liver function and poorer prognosis. Neurodevelopmental delay, and other subtle symptoms such as school problems or lethargy can reflect chronic hepatic encephalopathy. Spontaneous bacterial peritonitis should always be suspected in children with ascitis, abdominal pain and fever. Paracentesis reveals cloudy fluid with a neutrophil count of $>250/\text{ml}$. *Klebsiella*, *E. coli* or *strep. pneumoniae* predominant.

Question 20

The following are features found in intravascular haemolysis

- A- Red cell fragments
- B- Haemoglobinaemia
- C- Haemosiderinuria
- D- Haemoglobinuria
- E- Methaemoglobinaemia

Answer Question 20

A- True B- True C- True D- True E- True

All of the above are seen in intravascular haemolysis, which is the destruction of red cell within the circulation. The particular features of intravascular haemolysis are:

- *Haemoglobinaemia
- *Haemoglobinuria
- *Haemosiderinuria
- *Methaemoglobinaemia
- *Red cell fragments





Question 21

Features of homozygous sickle cell disease include

- A- Dactylitis
- B- Presentation usually during the first few months of life
- C- Increased incidence of Salmonella osteomyelitis
- D- Proliferative retinopathy
- E- Pigment gallstones

Answer Question 21

A- True B- False C- True D- True E- True

Homozygous sickle cell disease does not usually present in infancy, features include painful crises, haemolytic crises, acute sequestration and aplastic crises. Painful crises (vascular-occlusive) occur in the bones (most common). The other organ may also be involved including the kidney, liver, spleen, heart, brain and chest. Painful swollen fingers (Dactylitis) may be seen, with bony infarcts resulting in different length fingers. Other features include pigment gallstones, leg ulcers, salmonella osteomyelitis and proliferative retinopathy.

Question 22

A prolonged bleeding time is seen in

- A- Von Willebrand's disease
- B- Bernard-Soulier syndrome
- C- Glanzmann's disease
- D- Hermansky-Pudlak syndrome
- E- Haemophilia B

Answer Question 22

A- True B- True C- True D- True E- False

The bleeding time measures platelet plug formation in vivo, A prolonged bleeding time is seen in thrombocytopaenia and in platelet function disorders. These include:

- *Bernard-Soulier syndrome (Platelet adhesion defect, few large platelet)
- *Glanzmann's disease (failure of platelet aggregation due to a deficiency of membrane glycoproteins Hb and IIIa)
- *Hermansky-Pudlak syndrome (platelet function disorder and albinism)
- *Von Willebrand's disease (platelet adhesion abnormalities present)
- *The bleeding time is normal in Haemophilia A and B





Question 23

The following are causes of neonatal thrombocytopaenia

- A- Bernard-Soulier syndrome
- B- Congenital rubella infection
- C- Leukaemia
- D- Renal vein thrombosis
- E- Giant haemangioma

Answer Question 23

A- False B- True C- True D- True E- True

The causes of thrombocytopaenia in the neonatal period are many. They may be due to an increased consumption of platelets or decreased production.

Bernard-Soulier syndrome is a disorder of platelet function not numbers. Congenital infections can result in neonatal thrombocytopaenia, and the infant may have a purpuric rash. Neonatal leukaemia is a rare condition.

Renal vein thrombosis and giant haemangiomas result in platelet fall due to a consumptive coagulopathy.

Question 24

Osteosarcoma

- A- Is a small round cell neoplasm
- B- Has a predilection for the flat bones
- C- Is associated with retinoblastoma
- D- Is less common than Ewing's sarcoma
- E- X-ray findings include lytic lesions

Answer Question 24

A- False B- False C- True D- False E- False

Osteosarcoma is a spindle cell neoplasm (Ewing's tumour is a small cell tumour). It occurs mainly in the metaphyses of the long bones, particularly the proximal femur and the distal tibia. It is associated with retinoblastoma and Osteogenesis imperfecta. It is more common than Ewing's tumour.

The x-ray findings are sclerotic lesions and skip lesions (lytic lesions are seen in Ewing's tumours).





Question 25

In acute myeloid leukaemia (AML)

- A- Gum hypertrophy is a clinical feature of the M4 and M5 subtypes
- B- Fanconi's syndrome is associated
- C- Auer rods are always seen
- D- DIC is feature of the M3 (acute promyelocytic) subtype
- E- Retinoic acid may be used as initial therapy in the M2 subtype

Answer Question 25

A- True B- True C- False D- True E- False

Acute myeloid leukaemia (AML) has many subtypes classified by the French-American-British (FAB) system. Gum hypertrophy is seen particularly in M4 and M5 types and DIC in M3 type. The M3 type has a good prognosis and may be treated with retinoic acid as initial therapy. Fanconi anaemia is associated with AML, as is trisomy 21 and Bloom syndrome. Auer rods are not always seen, depending on the type of ALL.

Question 26

Bruton's X-linked agammaglobulinaemia

- A- Involves a mutation in the btk gene
- B- Is inherited as Autosomal recessive
- C- Is associated with unusual enteroviral infections
- D- A block is present in the maturation of pre-B cells
- E- Usually presents with pneumocystis carinii pneumonia

Answer Question 26

A- True B- False C- True D- True E- False

Bruton's X-linked agammaglobulinaemia is an x-linked recessive disorder of humoral immunity. The gene defect is a mutation in the btk gene which is on the X chromosome and results in an absence of tyrosine kinase. There is defective transformation of pre-B cells to B cells. The disease presents with recurrent bacterial infections and there may be unusual enteroviral infections (i.e. chronic encephalomyelitis). Pneumocystis carinii infection is seen in T cell immunodeficiency. Regular immunoglobulin infusions (3-4 weekly) are necessary throughout life.





Question 27

In Job's syndrome

- A- IgM counts are very high
- B- It is due to mutations in the intracellular kinase Jak3
- C- Eczema is a feature
- D- Small platelets are seen
- E- Recurrent staphylococcal skin abscesses occur

Answer Question 27

A- False B- False C- True D- False E- True

Job's syndrome (also known as Hyper IgE syndrome) is a syndrome of very high levels of IgE, eczema and recurrent staphylococcal abscesses in the skin, joints and lungs. Small platelets are seen in Wiskott-Aldrich syndrome. The mutations in Jak3 result in Autosomal recessive T-9, B+SCID

Question 28

The Haemophilus influenza b vaccine

- A- Is composed of the polysaccharide coat and a protein
- B- Provides protection against non-encapsulated organisms
- C- Protects against epiglottitis
- D- Is a live vaccine
- E- Is effective in the neonate

Answer Question 28

A- True B- False C- True D- False E- False

The haemophilus influenza b (Hib) vaccine is composed of the polysaccharide coat and a protein to make it antigenic. It provides protection to the encapsulated organism haemophilus influenza. This protects from acute epiglottitis and meningitis. It is not a live vaccine. It is not effective in the neonate because maternal Hib antibodies interfere with its effect.





Question 29

Mumps

- A- Is a DNA virus
- B- Is a common cause of viral meningitis
- C- Involves purely the submandibular gland in 20% of cases
- D- Is a cause of pancreatitis
- E- May cause myocarditis

Answer Question 29

A- False B- True C- False D- True E- True

Mumps virus is one of the paramyxoviruses which are RNA viruses. It is very common cause of viral meningitis.

It usually involves the parotid gland (60%) or both the parotid and submandibular. It is rare to be only involve the submandibular gland. It is a cause of both pancreatitis and myocarditis.

Question 30

Concerning malaria

- A- Plasmodium vivax infects only the old red blood cells
- B- Nephrotic syndrome can be seen in plasmodium malaria infection
- C- Plasmodium falciparum has a latent phase in the liver
- D- Sporozoites rupture to release merozoites in the blood
- E- Primaquine is needed for eradication in plasmodium malariae

Answer Question 30

A- False B- True C- False D- False E- False

Plasmodium vivax and P.ovale affect the young red blood cells. P.malaria affect the old blood cells. Plasmodium falciparum does not have a latent phase in the liver unlike P.vivax and P.ovale. Schizont rupture releases merozoites. Primaquine is not needed for liver eradication in P.malaria infection as there is no latent phase in the liver.





Question 31

Regarding leprosy

- A- The incubation period is 1-3 months
- B- The lepromin test is negative in Lepromatous leprosy
- C- The diagnosis is by culturing the organism in artificial media
- D- The clinical disease is dependant on the immune status of the individual
- E- Tuberculoid leprosy is seen in people with poor cell-mediated immunity

Answer Question 31

A- False B- True C- False D- True E- False

Leprosy is caused by Mycobacterium leprae. It is very slow growing and the incubation period is 3-5 years. The lepromin test is a measure of host resistance to disease, and is negative in Lepromatous leprosy where the host cell-mediated immunity is poor. The clinical features of the disease are dependent on the immune status of the host, and Tuberculoid leprosy is seen in people with a good immune response.

Question 32

In bacterial meningitis

- A- CSF glucose : plasma ratio is high
- B- Approximately one fifth of infants will present with seizures
- C- Subdural effusions are rare
- D- Kernig sign is almost always positive
- E- Rifampicin is given to contacts of pneumonia meningitis

Answer Question 32

A- False B- True C- False D- False E- False

CSF Glucose : Plasma ratio is low in bacterial meningitis. Subdural effusions occur in 10-15% of cases of bacterial meningitis.

Kernig sign is often negative in children, especially young infants.

Up to a fifth of infants present with seizures, and a further fifth will have seizures later on.

Rifampicin is given to contacts of N.meningitidis and H.influenzae meningitis, but not S.pneumoniae.





Question 33

In galactosaemia

- A- there is an incidence of 1 in 6000
- B- The enzyme deficiency is glucose phosphorylase
- C- There is an inability to metabolise galactose and lactose
- D- Diagnosis is by red blood cell enzyme assay
- E- Speech problems are almost inevitable even with therapy

Answer Question 33

A- False B- False C- True D- True E- True

Galactosaemia has an incidence of 1 in 60,000. The enzyme deficiency is of galactose-1-phosphate uridylyl transferase (GALT). This results in an inability to metabolise galactose (and thus also lactose, which is made of glucose+galactose).

Clinical features include presentation as a neonate with vomiting, hypoglycaemia, feeding difficulties, neurological features and liver disorder. Diagnosis is by enzyme assay in RBCs. Non-glucose reducing substances are seen in the urine when milk fed (i.e. Clinitest positive, Clinistix negative). The Guthrie test for galactosaemia.

Management involves a lactose and galactose free diet. Speech-Language problems and ovarian failure though, are almost inevitable even with therapy.

Question 34

The mitochondrial disorders

- A- May present at any age and by mode of inheritance
- B- Include Pearson's syndrome
- C- Features may include retinitis pigmentosa
- D- Include myoclonic epilepsy and ragged-red fibres (MERRF)
- E- Elevated free-floating blood lactate supports the diagnosis

Answer Question 34

A- True B- True C- True D- True E- True

The mitochondrial disorders may indeed present at any age and by any mode of inheritance. They are usually Autosomal recessive. They include MERRF, Pearson's syndrome, MELAS (mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes) syndrome and Leigh's syndrome.

Diagnosis is suggested by an elevated free-floating blood lactate in the absence of sepsis, hypoxia, poor tissue perfusion or other metabolic disorder which causes high lactate. Diagnosis is confirmed by enzyme analysis.

Features are many and variable, and they include retinitis pigmentosa.





Question 35

X-linked adrenoleucodystrophy

- A- Is a mitochondrial disorder
- B- May present as Addison's disease
- C- Involves accumulation of short chain fatty acids
- D- Involves progressive neuronal white matter degeneration
- E- Lorenzo's oil is curative

Answer Question 35

A- False B- True C- False D- True E- False

X-linked adrenoleucodystrophy is a peroxisomal disorder of very long chain fatty acid (VLCFA) oxidation. It results in progressive adrenal cortex and neuronal white matter degeneration, and the phenotype is variable. It may present with Addison's disease, or it may present with neurological features (behaviour disturbance, developmental regression, ataxia, seizures, spasticity). Diagnostic investigations include VLCFAs, adrenal cortical function tests, MRI brain scan and neuropsychiatric assessment.

Management is supportive with adrenal steroid replacement, anticonvulsants, NG or gastrostomy feeding. Supplementation with monounsaturated amino acids (Lorenzo's oil) may be beneficial but is not curative.

Question 36

In congenital Hypothyroidism

- A- Ectopic thyroid tissue is the commonest finding
- B- Dyshormonogenesis accounts for around 25%
- C- The incidence is around 1 in 6000
- D- May result from maternal hyperthyroidism
- E- Significant intellectual impairment is inevitable

Answer Question 36

A- True B- False C- False D- True E- False

Congenital hypothyroidism occurs with an incidence of around 1 in 4000. It is most commonly due to thyroid dysgenesis with ectopic thyroid tissue. Dyshormonogenesis accounts for only about 10% of cases. Maternal hyperthyroidism may result in congenital hypothyroidism through antibodies crossing the placenta, or from the antithyroid drugs. Significant intellectual impairment is avoidable if hormone therapy is instituted early.





Question 37

The following may result in diabetes insipidus

- A- Craniopharyngioma
- B- Carbamazepine
- C- Demeclocycline
- D- Neonatal listeriosis
- E- The DIDMOAD syndrome

Answer Question 37

A- True B- False C- True D- True E- True

The cause of diabetes insipidus may be separated into intracranial causes and nephrogenic causes. Craniopharyngioma and neonatal listeriosis are causes of intracranial diabetes insipidus. DIDMOAD syndrome stands for diabetes insipidus, diabetes mellitus, optic atrophy and deafness. Demeclocycline can cause nephrogenic diabetes insipidus. Carbamazepine is used in the treatment of nephrogenic diabetes insipidus.

Question 38

Craniopharyngioma

- A- Is one of the most common infratentorial tumours in childhood
- B- May present with tall stature
- C- May present with polyuria
- D- Arises from a remnant of the connection between Rathke's pouch and the oral cavity
- E- Is associated with calcification on the skull x-ray in about 20%

Answer Question 38

A- False B- False C- True D- True E- False

Craniopharyngioma is one of the most common supratentorial tumours in childhood (not infratentorial). It may present with the hormonal effects of hypopituitarism, including growth failure, polyuria (due to diabetes insipidus), hypothyroidism and adrenocortical insufficiency. It arises from a remnant of the connection between the Rathke's pouch and the oral cavity. In most cases there is calcification on the skull x-ray.





Question 39

In familial hypophosphataemic rickets

- A- There is defective distal renal tubule reabsorption of phosphate
- B- Levels of 1,25 dihydroxycholecalciferol are normal
- C- Parathyroid hormone level are raised
- D- Plasma calcium levels are high
- E- The inheritance is Autosomal dominant

Answer Question 39

A- False B- False C- False D- False E- False

In familial hypophosphataemic rickets (also known as vitamin D-resistant rickets), there is defective proximal renal tubular reabsorption of phosphate, and reduced synthesis of 1,25-hydroxycholecalciferol. Plasma levels of PTH are normal, and calcium is normal or low. Plasma phosphate levels are low. The inheritance is x-linked dominant.

Question 40

Children high on the autistic spectrum

- A- Demonstrate repetitive behaviour
- B- Have distinctive abnormal findings on the EEG
- C- Have delayed motor milestones
- D- Have a good emotional contact with the mother but not with any strangers
- E- Have normal speech development

Answer Question 40

A- True B- False C- False D- False E- False

Children with autism demonstrate repetitive behaviour, have poor eye contact with others, and have a very poor emotional bond with both their mother and other people. The EEG is usually normal. They have normal motor development, but their speech is delayed with difficulty understanding language.





Question 41

Febrile convulsion

- A- Occur in 8 – 10% of children
- B- Are more common in male than females
- C- Typically occur between 6 months and 6 years
- D- Are not associated with increased risk of later development of epilepsy
- E- Are usually associated with a positive family history

Answer Question 41

A- False B- True C- True D- False E- False

Febrile convulsion occur in around 2-5% of children and are more common in males. They occur between 6 months and 6 years. They are associated with an increased risk of later development of epilepsy, particularly if atypical and there is a family history of epilepsy. There is a positive family history of febrile convulsion in around 30% of cases.

Question 42

Features of late infantile Batten disease include

- A- Acute intermittent ataxia
- B- Developmental regression
- C- Diagnosis on rectal biopsy
- D- X-linked recessive inheritance
- E- Retinitis pigmentosa

Answer Question 42

A- False B- True C- True D- False E- True

Late infantile Batten's disease (Ceroid Lipofuscinosis) is an Autosomal recessive condition. The features are normal early development then developmental regression from around 2-5 years. There is a chronic ataxia and retinitis pigmentosa can occur. Typical neurological features are seen on histology of rectal biopsy.





Question 43

In raised intracranial pressure

- A- Drowsiness is seen with a rapidly increasing pressure
- B- Convulsions are a common presentation
- C- The headache is worse on movement
- D- There may be a sixth nerve palsy
- E- Signs take longer to develop in young children than in adults

Answer Question 43

A- True B- False C- True D- True E- True

Raised intracranial pressure may develop insidiously or rapidly and drowsiness is a feature of a rapidly rising pressure. Convulsions are an unusual presentation in children. The headache is worse on lying down and in the morning. There may be a sixth nerve palsy (a false localising sign). Signs take longer to develop in young children as the cranial sutures can widen.

Question 44

Slipped upper femoral epiphysis

- A- Is seen in children age 5-10 years most commonly
- B- Is associated with hypothyroidism
- C- May present with knee pain
- D- Is seen on hip x-ray with a narrowing of the growth plate
- E- Shows decreased external rotation of the hip on examination

Answer Question 44

A- False B- True C- True D- False E- False

Slipped upper femoral epiphysis is seen most commonly in adolescent. It is associated with hypothyroidism and other pituitary dysfunction. It may present with referred pain from the knee. The hip x-ray shows widening of the growth plate as the head slip's off the neck.

Examination reveals decreased internal rotation of the hip, and the hip is chronically externally rotated.





Question 45

The following concerning antimalarials are true

- A- Chloroquine is always contraindicated in G6PD
- B- Proguanil inhibits Folate production
- C- Mefloquine causes neuropsychiatric symptoms in up to 10% of patients
- D- Pyrimethamine resistance is low
- E- Long term Chloroquine is associated with cataract

Answer Question 45

A- False B- True C- True D- False E- False

Chloroquine may be given in G6PD deficiency though haemolysis can occur in some forms of the disease, dapson and sulphur containing drugs should be avoided. Proguanil acts by inhibiting folate production, and the parasitic cells can not utilise tetrahydrofolate from external sources unlike mammalian cells. Pyrimethamine resistance is high and it is no longer recommended as prophylaxis for travellers. Long term Chloroquine is associated with corneal opacities and retinal damage.

Question 46

Doxorubicin

- A- Is an anthracycline
- B- Is more effective against solid tumours than daunorubicin
- C- Is cardiotoxic to all patients
- D- Binds to DNA
- E- Is safe in patients with a history of shingles

Answer Question 46

A- True B- True C- True D- True E- False

Doxorubicin is a naturally occurring anthracycline and derived from streptomyces. It is effective against solid tumours and is part of the regime of many solid tumours. It induces free radical formation which cardiac cells specifically can not deal with; the clinical effects though are variable. Doxorubicin binds to DNA and alters the DNA helix shape, thus inhibiting DNA polymerase. There is a risk of severe infection with disseminated zoster in patients with a history of shingles.





Question 47

Liver enzyme inducers include

- A- Carbamazepine
- B- Rifampicin
- C- Brussel sprouts
- D- Phenytoin
- E- Tobacco smoke

Answer Question 47

A- True B- True C- True D- True E- True

All of these are liver enzyme inducers, Brussel sprouts and barbecued meat both acts as liver enzyme inducers. Heavy smoking and drinking may account for a failure to respond to a normal drug dose.

Question 48

The following are true of salicylate poisoning

- A- Hypokalaemia is a feature
- B- Hypoventilation is an early feature
- C- Vasodilatation is a feature
- D- Coma indicate sever poisoning
- E- Respiratory alkalosis is seen in children

Answer Question 48

A- True B- False C- True D- True E- True

Hypoventilation is a late feature of salicylate poisoning, and together with coma indicate sever poisoning. The acid-base disturbances are complex and both acidosis and alkalosis may occur. Salicylates have a direct stimulant effect on the respiratory centre resulting in hyperventilation and a respiratory alkalosis seen early on, though this stage may not be apparent in young infants.





Question 49

Mollusca contagiosa

- A- Is caused by a pox virus
- B- Is seen as a pearly papule with a central umbilicus
- C- Spontaneous resolution is usual by 6 weeks
- D- May be disseminated in a child with atopic eczema
- E- May be treated with cryotherapy

Answer Question 49

A- True B- True C- False D- True E- True

Mollusca contagiosa is a common infection among school children caused by a pox virus. It presents as pearly papules with a central umbilicus. Spontaneous resolution usually occurs within 6-9 months, though the lesions can last for years. Disseminated infection may be seen in children with atopic eczema and in the immunosuppressed. No treatment is usually given as this can result in scarring, however, cryotherapy is a treatment option if necessary.

Question 50

Reiter's syndrome

- A- Is associated with campylobacter gastroenteritis
- B- Responds well to antibiotics
- C- Keratoderma blenorrhagica may be a feature
- D- Usually resolves within a year of onset
- E- Does not become chronic

Answer Question 50

A- True B- False C- True D- True E- False

Reiter's syndrome is an acute reactive arthritis. It follows gastrointestinal infection (including campylobacter, shigella, yersinia, salmonella) or venereal infection (Chlamydia or NSU).

It has a high male preponderance, and is associated with HLA-B27 (80%).

Features include a lower limb arthritis, which may become chronic, ocular inflammation and a sterile urethritis. Other features are Keratoderma blenorrhagica, plantar fasciitis, enthesopathy, nail dystrophy and mouth ulceration. Treatment is with antibiotics, physiotherapy and NSAIDs.





Question 51

Regarding Kawasaki disease

- A- It is a Polyarteritis
- B- A fever of $>37.5^{\circ}\text{C}$ should be present for 5 days to make the diagnosis
- C- Untreated, nearly 2% of children will develop coronary artery aneurysms
- D- Intravenous immunoglobulin should be given within 3 weeks of onset of disease
- E- A marked thrombocytopenia is seen in the second and third week of disease

Answer Question 51

A- True B- False C- False D- False E- False

Kawasaki disease is an infantile polyarthritis. The diagnostic criteria are a fever of $>38.5^{\circ}\text{C}$ for >5 days together with 4 of the following features:

- *Bilateral non-purulent conjunctivitis.
- *Oral mucosal changes.
- *Cervical lymphadenopathy with one node $>1.5\text{ cm}$.
- *Involvement of hands and feet with erythema, swelling or peeling of the palms and soles.
- *Rash (polymorphous).

The child is usually extremely irritable, with cough or coryzal symptoms, and may have watery diarrhea.

Cardiac complications are significant cause of morbidity: coronary artery aneurysms occur in up to 20% of children who are not treated.

A thrombocythaemia is seen in the second and third weeks, and a high WCC and anaemia may be seen.

Management is with high dose intravenous immunoglobulin over 12 hours, which should be given within 10 days of disease onset. Aspirin is given for 6 weeks or until the coronary aneurysms are gone, which is assessed by echocardiogram at follow-up.





Question 52

Syndromes involving absent radii include

- A- Stickler's dysplasia
- B- Holt-Oram syndrome
- C- Ellis-Van Creveld syndrome
- D- Fanconi anaemia
- E- VATER syndrome

Answer Question 52

A- False B- True C- False D- True E- True

Syndromes involving absent radii include:

- Holt-Oram syndrome
- Fanconi anaemia
- VATER syndrome
- TAR syndrome
- Absent thumbs

Question 53

Features of Noonan syndrome include

- A- Peripheral pulmonary stenosis
- B- Ventricular septal defect
- C- Ptosis
- D- Mental retardation
- E- Autosomal dominant inheritance

Answer Question 53

A- True B- True C- True D- True E- True

Noonan syndrome is inherited in an Autosomal dominant fashion, though most cases are sporadic. The appearance may be similar to Turner's syndrome and features include:

- *Mental retardation.
- *Ptosis.
- *Epicanthic folds.
- *Hypertelorism.
- *Down-slanting palpebral fissures.
- *Cardiac defects: these include valvular pulmonary stenosis, PDA, VSD, and peripheral pulmonary stenosis.





Question 54

In embryological development of the lung

- A- The lung bud is an outgrowth of the midgut
- B- The respiratory bronchioles develop from 22 weeks gestation
- C- The lung bud derives from mesodermal tissue
- D- Type I pneumocytes produce surfactant
- E- Surfactant production can be detected by 23 weeks gestation

Answer Question 54

A- False B- False C- False D- False E- True

The lung bud is an outgrowth from the foregut and derived from endodermal tissue. The respiratory bronchioles develop from about 17 weeks gestation and surfactant production can be detected by about 23 weeks gestation. The type II pneumocytes produce surfactant.

Question 55

Multifactorial inheritance

- A- Is seen in the inheritance of pyloric stenosis
- B- Involves both genetic and environmental factors
- C- Means that the inheritance risk for relatives can not be estimated
- D- Operates in the inheritance of neural tube defects
- E- Risks for multifactorial inheritance increase if more family members are affected

Answer Question 55

A- True B- True C- False D- True E- True

Multifactorial inheritance involves both genetic and environmental factors. Many diseases have multifactorial inheritance including cleft lip and palate, pyloric stenosis, neural tube defects, club foot and congenital hip dislocation. Empirical recurrence risk are based on studies of large collections of families. The risk increases if more family members are affected, if the disease has more severe expression and if the affected case is a member of the less commonly affected sex.





Question 56

The following are true

- A-A single blind trial indicates that neither the patient nor the assessor know which group a patient has been assigned to
- B-The Chi-squared test is carried out using percentages
- C-Randomisation will help eliminate selection bias
- D-The median is the value which occurs most often
- E-The incidence of a disorder is the number of the population suffering from a disorder at one point in time

Answer Question 56

A- False B- False C- True D- False E- False

A single blind trial is one in which either the patient or the assessor, but not both, does not know which group the patient has been assigned to.

The Chi-squared test is carried out using numerical data only and not percentages.

Randomisation will help eliminate selection bias because it ensures all patients should have an equal chance of being assigned to each group.

The median is the value which divides the range of values in to two equal parts. The incidence of a disorder is the number of new cases occurring over a set period of time....

Question 57

Human breast milk

- A- Contains less carbohydrate than cow's milk
- B- Contains less iron than cow's milk
- C- Is associated with an increased risk of haemorrhagic disease of the newborn
- D- Has a higher casein to whey ratio than cow's milk
- E- Has more protein than cow's milk

Answer Question 57

A- False B- False C- True D- False E- False

Human breast milk contains more carbohydrate, more iron, less protein than cow's milk, and it has a lower casein to whey ratio.

It is a poor source of vitamin K and is therefore a risk factor for haemorrhagic disease of the newborn.





Question 58

The following conditions are associated with delayed closure of the anterior fontanelle

- A- Achondroplasia
- B- Apert syndrome
- C- Hyperthyroidism
- D- Progeria
- E- Malnutrition

Answer Question 58

A- True B- True C- False D- True E- True

Conditions associated with premature closure of the fontanelle include many skeletal disorders, such as Achondroplasia, Apert syndrome, and Osteogenesis imperfecta. Progeria is also associated with delayed closure of the fontanelle. Malnutrition may result in delayed closure. Other conditions associated with delayed closure include Trisomy 13, 18 and 21.

Hyperthyroidism is associated with premature closure, and hypothyroidism with delayed closure of the fontanelle.

Question 59

The following may result in neonatal hypocalcaemia

- A- Infant of a diabetic mother
- B- Birth asphyxia
- C- Infant fed on high-phosphate cow's formula
- D- Hypermagnesaemia
- E- Neonatal hyperparathyroidism

Answer Question 59

A- True B- True C- True D- False E- False

Neonatal hypocalcaemia is seen in infants of diabetic mothers, and in infants with birth asphyxia. Infants fed on cow's milk formula can develop hypocalcaemia because this milk is very high in phosphate which the neonatal kidney can not manage to excrete. This results in increased bone deposition of calcium, decreased 1,25-dihydroxy vitamin D levels and hypocalcaemia.

Hypomagnesaemia and Neonatal hypoparathyroidism will result in hypocalcaemia.





Question 60

In twin pregnancy

- A- In twin-twin transfusion the plethoric twin is at greater risk
- B- There is an increased risk of pre-eclampsia
- C- There is an increased risk of placenta praevia
- D- Dizygotic twins are at great risk of complications than monozygotic twins
- E- Monozygotic twins are always monochorionic

Answer Question 60

A- True B- True C- True D- False E- False

In twin-twin transfusion the larger plethoric twin is the one at greater risk of a number of complications. There is an increased risk of almost everything in twin pregnancy, including pre-eclampsia and placenta praevia.

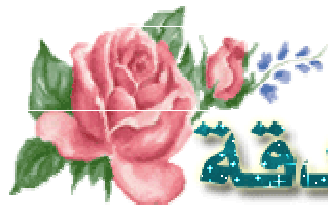
Monozygotic twins are at greater risk than Dizygotic because if the chorion is shared there are risks of vascular anastomosis with twin-twin transfusion and cord entangling. Monozygotic twins, may however, be dichorionic.

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الكلمة الطيبة صدقة

