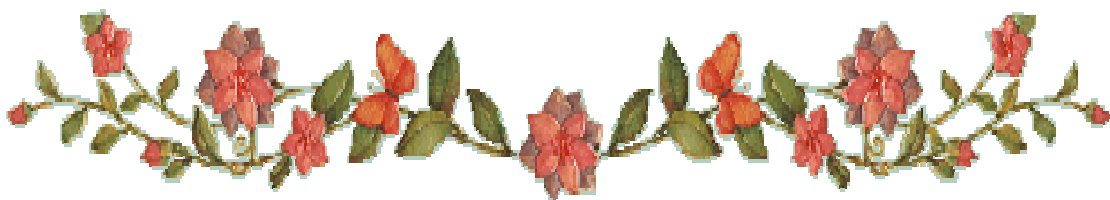




*Pediatric*  
*Questions & Answers*  
*Group 2-2008*





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### Question 1

#### In a patient with a small VSD

- A- symptoms are usually present at birth
- B- The risk of endocarditis is an indication for closure of the defect.
- C- The ECG is usually normal
- D- If the systolic murmur is loud the prognosis is worse
- E- 10% will close spontaneously in the first few years of life.

### Answer Question 1

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

A small VSD is often asymptomatic and most will close spontaneously during the first few years of life. The risk of endocarditis is not an indication for closure, though, prophylaxis will be needed for certain procedures. The small VSDs have shorter louder systolic murmurs. A mid-diastolic apical murmur is heard with large defects. The ECG is normal and may show LVH if the murmur is large. If pulmonary hypertension develops, then RVH is seen on the ECG.

Surgical repair is indicated if there are severe symptoms with failure to thrive, if pulmonary hypertension develops, if aortic regurgitation develops, or if there is persistent significant shunting over 10 years of age.

### Question 2

#### The following are features of a Supraventricular tachycardia.

- A- The tachycardia responds to adenosine.
- B- Fusion beats are present.
- C- There is AV dissociation.
- D- The heart rate is always regular.
- E- Capture beats are present.

### Answer Question 2

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

A supraventricular tachycardia (SVT) can be hard to distinguish from a ventricular tachycardia (VT).

Fusion and capture beats are seen in ventricular tachycardia.

AV association is present in SVT (Dissociation in VT).

The heart rate may not be regular in SVT. SVT responds to adenosine.

If there is uncertainty about the nature of the arrhythmia (SVT or VT) then it should be treated as VT..





### *Question 3*

**Peripheral pulmonary stenosis is seen in**

- A- Alagille syndrome
- B- Marfan's syndrome
- C- Klinefelter's syndrome
- D- Congenital rubella syndrome
- E- William's syndrome

### *Answer Question 3*

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

Peripheral pulmonary stenosis is seen in William's syndrome, Congenital rubella syndrome, and Alagille syndrome.

### *Question 4*

**A right sided aortic arch is seen with**

- A-Ebstein's anomaly
- B-Tetralogy of Fallot
- C-Congenital vascular ring
- D-Pulmonary atresia
- E-Truncus arteriosus

### *Answer Question 4*

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

A right sided aortic arch is seen in Fallot's tetralogy, truncus arteriosus, pulmonary atresia and commonly with a congenital vascular ring. It may also be present with no cardiac abnormality.





### Question 5

#### In *Pneumocystis carinii* pneumonia

- A- Presentation in infants is usually with acute onset of dyspnea
- B- Investigation may require bronchoalveolar lavage
- C- Septrin may cause a neutropaenia
- D- The chest x-ray is normal on presentation in around a quarter of cases
- E- Post-infection prophylaxis with Septrin is sometimes necessary in the immunocompromised.

### Answer Question 5

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

*Pneumocystis carinii* pneumonia in infants usually presents with an insidious onset of dyspnea, tachypnea, cough and fever at around 3 months in the immunocompromised infant. Septrin therapy can cause a neutropaenia, and also a rash. Bronchoalveolar lavage is necessary for diagnosis if sputum can not be obtained otherwise, hypoxia is a cardinal feature, and is often severe despite normal chest auscultation. The chest x-ray is normal in about 25% at presentation. Otherwise it shows perihilar shadowing “ butterfly rash ” or a ground glass appearance with air bronchograms.

Post-infection prophylaxis with Septrin or pentamidine is recommended in the immunocompromised.

### Question 6

#### Cystic Adenomatoid malformation (CAM) of the lung

- A- Is the most common congenital malformation of the lung
- B- Rarely results in midline shift with compression of the opposite lung
- C- May present with recurrent chest infection
- D- Is associated with Turner's syndrome
- E- Is managed with surgical resection.

### Answer Question 6

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

Cystic Adenomatoid malformation (CAM) of the lung is the second most common congenital lung malformation (congenital lobar emphysema being the most common). It usually causes a midline shift and compression of the opposite lung. It presents most commonly with neonatal respiratory distress, though may present with recurrent chest infections. Management is surgical with resection. There is no known association with Turner's syndrome.





## Question 7

### Regarding cystic fibrosis

- A- Nasal polyps in childhood and adolescence are virtually pathognomonic of cystic fibrosis.
- B- It may present in infancy with severe gastro-oesophageal reflux.
- C- The commonest cause of a respiratory exacerbation during infancy is Haemophilus influenza.
- D- Flucloxacillin does not interfere with the sweat test.
- E- Treatment of allergic bronchopulmonary aspergillosis is with itraconazole as first line therapy.

## Answer Question 7

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

Nasal polyps in adults are more likely to signify aspirin sensitive asthma.

Staphylococcus aureus is the most common pathogen in infancy .

After the age of 2 years the incidence of pseudomonas slowly rises.

Allergic bronchopulmonary aspergillosis is an allergic response to aspergillus, and therefore the treatment is steroids. The diagnosis is made by a combination of cough and wheeze, patch shadowing on chest x-ray and a high specific IgE to Aspergillus.

## Question 8

### In a child who appears to have malabsorption

- A- Anti-endomysial antibody is 90.5% sensitive for Coeliac disease.
- B- Faecal elastase is the most practically useful test for pancreatic insufficiency.
- C- Giardiasis may be diagnosed by faecal examination in 70% of cases.
- D- A small bowel biopsy is necessary for the diagnosis of a beta-lipoproteinaemia.
- E- Primary lactose deficiency is a common cause.

## Answer Question 8

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

Anti-endomysial antibody is an IgA and as 1-4% of the general population have low IgA there may be a FALSE negative result in these individuals. A maximum pick up rate of Giardia in the stool of 20% can be expected, and hence a trial of metronidazole for 5-7 days may be a better diagnostic tool. Primary lactase deficiency is rare, whereas post-gastroenteritis secondary lactase deficiency is not uncommon. Late onset congenital lactase deficiency occurs around the age of 10-14 especially in those of Mediterranean origin.





### *Question 9*

**Features of a non-organic origin for recurrent abdominal pain in children and adolescents are:**

- A- Early morning waking with pain
- B- Absence of dysuria.
- C- Predominance in girls.
- D- A family history of atypical migraine.
- E- Negative correlation with *Helicobacter pylori* serological positivity.

### *Answer Question 9*

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

Any nocturnal waking with pain must be investigated for an organic cause. UTIs can occur without symptoms and an MSU is necessary in the majority of children with abdominal pain even if a psychogenic origin is suspected. Abdominal migraine occurs and usually has a preceding family history of classical migraine. There is no correlation, positive or negative between *H. pylori* serology and RAP of non-organic origin

### *Question 10*

**Recognised pathology and complications of Crohn's include:**

- A- Toxic megacolon
- B- Increased presence of megakaryocytes.
- C- Calcium oxalate renal calculi.
- D- Caseating granulomas.
- E- Eventual diminished adult height if testicular volume is 5-10ml at 17 years of age.

### *Answer Question 10*

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

Toxic megacolon is a feature of ulcerative colitis. Terminal ileal involvement may cause a low vitamin B12. Non-caseating granulomas occur. Until testicular volume is 20 ml there is still potential for increase in height. The warning area where urgent attention is necessary to treat the affected bowel to allow the pubertal growth spurt to occur is reached when testicular volume reaches 10 ml.







### Question 11

#### Regarding micronutrient and vitamin deficiencies:

- A- Dermatitis, dementia, and diarrhea occurs with a deficiency of niacin (nicotinamide).
- B- Symmetrical polyneuropathy occurs with thiamine deficiency.
- C- Raised S-T segments on ECG can occur with vitamin A deficiency.
- D- Selenium deficiency occurs within 6 weeks of commencement of total parenteral nutrition.
- E- Vitamin A is found mainly in fish when a child is on a dairy-free diet.

### Answer Question 11

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

Selenium stores are sufficient to account for requirements for 6 months when on a selenium-free diet (modern TPN will have selenium added to it), and deficiency can cause cardiomyopathy not pericarditis. Eggs, Liver, and green vegetables are good sources of vitamin A.

### Question 12

#### Atrial natriuretic peptide:

- A- Increases glomerular filtration rate.
- B- Decreases blood pressure.
- C- Decreases renin-angiotensin-aldosterone system action.
- D- Is secreted from the cardiac atria in response to increased stretch, increased pressure and increased osmolarity.
- E- Causes peripheral vasoconstriction.

### Answer Question 12

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

Atrial natriuretic peptide also increases sodium and water excretion. It has no effect on vasoconstriction.







### Question 13

**Low complement levels are a finding in**

- A- Recurrent severe pyelonephritis.
- B- Mesangiocapillary glomerulonephritis.
- C- Post-streptococcal glomerulonephritis.
- D- Congenital Nephrotic syndrome.
- E- Focal segmental glomerulonephritis.

### Answer Question 13

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

In type I mesangiocapillary GN a low C3 and normal C4 are classically observed with subendothelial immune complex deposition and splitting of the basement membrane.

Type II results in mesangial cell proliferation and intramembranous immune complex deposition. Post-streptococcal GN (e.g. diffuse GN) may have low C3 and normal C4, focal segmental, and rapidly progressive GN do not classically have complement decrease. Similarly membranous and minimal change do not have a low complement as a common feature.

### Question 14

**Proximal renal tubular acidosis is associated with.**

- A- Proximal tubular bicarbonate secretion
- B- Rickets.
- C- Aminoaciduria.
- D- Hypochloraemia.
- E- Interstitial nephritis

### Answer Question 14

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

Proximal RTA is due to proximal tubule bicarbonate reabsorption, whereas distal RTA is due to distal tubule failure to excrete hydrogen ions. Rickets and aminoaciduria occur when associated with Fanconi's syndrome (Type II proximal RTA). Hyperchloraemia, Hypokalaemia, low serum bicarbonate, and a metabolic acidosis with urine which can be acidified below pH 5.5, unlike distal RTA where the urine can not be acidified below pH 5.8. secondary causes of distal RTA include interstitial nephritis, obstructive nephropathy, and pyelonephritis.





### *Question 15*

**Renal malformations occur in the following**

- A- Crohn's disease.
- B- Ulcerative colitis with positive pANCA.
- C- Tuberous sclerosis.
- D- Hemihypertrophy.
- E- DiGeorge syndrome.

### *Answer Question 15*

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

In some cases of UC, pANCA positivity may point towards multiple organ vasculitis and angiogram will reveal microaneurysms in the distribution of inferior mesenteric artery, renal bed, other visceral arterial supplies and even coronary arteries. Hamartomas or polycystic kidneys are associated with tuberous sclerosis. Hemihypertrophy may be associated with renal hyperplasia or Wilms' tumours.

### *Question 16*

**Biliary atresia is characterised by**

- A- An absence of a gallbladder on fasting ultrasound.
- B- Biliary duct proliferation on liver biopsy.
- C- Poor uptake of radioisotope in to the liver after pre-administration of phenobarbitone for 5 days.
- D- Facial dysmorphism with Hypertelorism, deep-set eyes and a small mandible.
- E- Conjugated hyperbilirubinaemia in the first 24 hours of life.

### *Answer Question 16*

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

Liver biopsy usually distinguishes between neonatal hepatitis and biliary atresia but the two can still present with similar histological hepatic features-biliary duct hyperplasia is usually seen in biliary atresia and giant cells are typical of neonatal hepatitis. Phenobarbitone pre-administration prior to a liver isotope scan increases the likelihood of hepatobiliary excretion in neonatal hepatitis but not biliary atresia-it has no effect on uptake in either condition. If facial dysmorphism is seen with so-called "pinched facies" of Hypertelorism, deep-set eyes, small mandible and a long nose then arteriohepatic dysplasia, or Alagille's syndrome, should be suspected in the presence of jaundice, biliary atresia is not characterised by jaundice in the first day of life as this is much more likely to be due to a haemolytic cause.





### Question 17

**The following are true of hepatitis B**

- A- Interferon alpha may seroconvert approximately 40% of infected children.
- B- HBsAB indicates a carrier state.
- C- 5-10% of infected children will develop fulminate hepatic failure.
- D- Hepatitis D virus can only occur in the presence of hepatitis B virus.
- E- HBeAB indicates high infectivity.

### Answer Question 17

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

After a 6 months course of 3-5 MIU/kg 3 times a week up to 40% of children will seroconvert to HBsAb and , where needed, HbsAg indicates carrier state, HBeAg indicates high infectivity, and HBsAb/HBeAb indicate seroconversion. In most studies no more than 1% of children go on to develop Fulminant liver failure, the vast majority recovering without sequelae. 10% develop a chronic carrier state, of whom 10-30% remain asymptomatic and 70-90% develop cirrhosis – both situation can lead to hepatocellular carcinoma. HDV or delta virus exists sloly in the HbsAg and is an incomplete RNA particle.

### Question 18

**In children with primary hepatic tumours**

- A- Hepatocellular carcinoma is commoner under the age of 4 than Hepatoblastoma.
- B- Abdominal pain will be the presenting feature in 90%.
- C- Jaundice occurs in less than 10%.
- D- Plain abdominal x-ray will demonstrate calcification in 40-50% of hepatocellular carcinomas.
- E- Hepatoblastoma has a well-established link with Beckwith-Weidemann syndrome and hemihypertrophy.

### Answer Question 18

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

Most hepatoblastomas occur under 18 months of age and HCC is commoner in older childhood. Short arm of chromosome 11 is implicated in the genetic etiology of Hepatoblastoma, and is associated with other embryonal tumours such as Wilm's. conversely, HCC seems to be associated with environmental factors. An abdominal mass will be present in 50-60% of HCCs and 70% of hepatoblastomas, but pain only occurs in 10-20%. Weight loss and anorexia are similarly uncommon 20%, and jaundice only appears in 7-10% of cases. MRI is the investigation of choice with further imaging of the vascularity by hepatic angiography if required. Calcification is not a feature.





### *Question 19*

#### **In paracetamol-induced hepatotoxicity in childhood**

- A- Hypoglycaemia is the commonest presenting feature.
- B- Concomitant ingestion of enzyme-inducers such as anti-convulsant drugs may increase risk of hepatotoxicity.
- C- Treatment with N-acetyl cysteine should be delayed until paracetamol levels are known.
- D- Subsequent autoimmune hepatitis is commoner than in the general population.
- E- A type IV systemic hypersensitivity reaction plays a part in the liver damage.

### *Answer Question 19*

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

Although hypoglycaemia may occur in fulminant liver failure, the commonest presentations are with symptoms such as nausea, anorexia and vomiting. Then after 24-48 hours, right upper quadrant pain, followed by signs of overt hepatic injury at day 2-4. normally it is conjugated to sulphate and glucuronide, but if this pathway is overwhelmed then the cytochrome P450 inducible system takes over and rapidly depletes the glutathione responsible for conjugating the toxic metabolites of this pathway from paracetamol. Hence enzyme induction will hasten toxicity. Paracetamol hepatotoxicity is a dose-dependent condition not reliant on hypersensitivity.





### Question 20

**The following are true regarding sideroblastic anaemia**

- A- It occurs as an X-linked recessive disease.
- B- It is seen in lead poisoning.
- C- A dimorphic blood film is often present.
- D- Pyridoxine may be used in therapy.
- E- Ring sideroblasts are seen in the marrow.

### Answer Question 20

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

In sideroblastic anaemia, hypochromic cells are seen in the peripheral blood, and ring sideroblasts are seen in the marrow with increased marrow iron (visible on Perl's reaction).

An inherited disease exists, which is x-linked recessive. Acquired disease may be primary (myelodysplasia FAB type 2) or secondary. Secondary disease is seen in malignant disease of the marrow, certain drugs (e.g. isoniazid), lead poisoning (basophilic stippling occurs), and haemolytic anaemia. The blood film shows microcytic, hypochromic cells, and is often dimorphic. The bone marrow shows erythroblasts with a ring of iron granules in them, and increased iron deposition.

Management involves removing any treatable cause. Pyridoxine therapy may help, particularly in inherited disease. Folate therapy is given if deficiency is present. Repeated blood transfusion may be necessary.

### Question 21

**In Diamond-Blackfan syndrome**

- A- A neutropaenia may be present.
- B- Presentation is usually in late childhood.
- C- Erythropoietin levels are raised in the blood.
- D- Triphalangeal thumbs are seen.
- E- The blood film shows a microcytic anaemia.

### Answer Question 21

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

Diamond-Blackfan syndrome is an Autosomal recessive condition. It is a red cell aplasia and present as a severe anaemia by 2-6 months of age. A neutropenia and a thrombocytosis may be present initially. The blood film shows a macrocytic anaemia with a young red cell population and reduced red cells. There are reduced red cell precursors in the bone marrow. Other abnormalities seen include triphalangeal thumbs, and dysmorphic facies.





### Question 22

#### Regarding idiopathic thrombocytopaenic purpura

- A- It is associated with Epstein Barr infection.
- B- Antiplatelet IgG antibodies are seen.
- C- It usually occurs during a viral infection.
- D- Chronic disease occurs in about a third of cases.
- E- Transfused platelets are quickly destroyed.

### Answer Question 22

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

idiopathic thrombocytopaenic purpura (ITP) is common in children. It usually occurs 1-4 weeks after a viral infection such as EBV, VZV and measles.

Clinical features are of bleeding with petechiae, bruises and mucosal bleeding. Intracranial haemorrhage occurs but is rare. Anti-platelet antibodies (both IgG and IgM) are seen. Most cases will resolve spontaneously in children, with only 5-10% becoming chronic.

Platelet transfusion are only given in emergency as they are quickly destroyed.

### Question 23

#### Causes of splenomegaly include.

- A- Osteopetrosis.
- B- Christmas disease.
- C- Diamond-Blackfan syndrome.
- D- Brucellosis.
- E- Neimann-Pick disease.

### Answer Question 23

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

Many causes of splenomegaly (useful to remember particularly for the clinical exam) may be classified into subgroups:

- \*Infectious causes e.g. EBV, subacute bacterial endocarditis, and brucellosis.
- \*Extramedullary haemopoiesis e.g. haemolytic anaemias, haemoglobinopathies, osteopetrosis.
- \*Congestion e.g. portal hypertension.
- \*Neoplastic conditions e.g. leukaemia.
- \*Storage diseases e.g. Neimann-Pick disease, Gaucher's disease, LCH, mucopolysaccharidoses.
- \*Other systemic diseases e.g. Amyloidosis, SLE.
- \*Massive splenomegaly is classically seen in malaria, kala-Azar, CML and myelofibrosis.





### Question 24

**The following are associated with Wilm's tumour**

- A- Mental retardation.
- B- Neurofibromatosis.
- C- Prader-Willi syndrome.
- D- Hemiplegia.
- E- Aniridia.

### Answer Question 24

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

Wilm's tumour is associated with neurofibromatosis, hemihypertrophy, aniridia, genitourinary anomalies and Beckwith-Weidemann syndrome.

### Question 25

**The following are associated with a good prognosis in acute lymphoblastic leukaemia (ALL).**

- A- Male sex.
- B- Age below 2 years at presentation.
- C- Hyperdiploidy.
- D- Translocation 4:11.
- E- Initial WCC low.

### Answer Question 25

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

The factors associated with a good prognosis in ALL are:

- \*Female sex.
- \*Low initial WCC.
- \*Age 2-10 years.
- \*Less than 4 weeks to initial remission.
- \*Translocation 12:21.
- \*c-ALL

The other features in the question are associated with a poor prognosis.







### *Question 26*

#### **Regarding immunoglobulins.**

- A- IgA is present in adult levels at birth.
- B- IgG levels fall in the months after birth.
- C- IgG is present in adult levels at birth.
- D- IgM reaches adult levels by puberty.
- E- Raised IgM levels at birth are seen in intra-uterine infection.

### *Answer Question 26*

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

IgG is present at adult levels at birth due to placental transfer of the immunoglobulin. The levels fall off during the first few months and reach lowest levels at 3-6 months before climbing slowly to reach adult levels by 5-6 years. This may result in transient hypogammaglobulinaemia of infancy. IgA is absent at birth and levels slowly rise to reach adult levels by puberty. IgM is also absent at birth with adult levels being reached by about 1 year of age. If there are increased levels of IgM at birth, this indicate intra-uterine infection.

### *Question 27*

#### **Regarding ataxia telangiectasia**

- A- Cell mediated immunity is impaired.
- B- IgA levels are normal.
- C- There is a sensitivity to ionising radiation.
- D- There are mutations in the ATM gene.
- E- Alpha-fetoprotein (AFP) is elevated.

### *Answer Question 27*

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

Ataxia telangiectasia involves both impaired cell mediated immunity and impaired antibody production. In particular, IgA is very low. IgE, IgG2 and IgG4 are also low. There is a defect in DNA repair, and an extreme sensitivity to ionising radiation. The genetic defect involves mutations in the ATM gene on chromosome 11. Both serum AFP and CEA are elevated.





### Question 28

**The following vaccines should be avoided in a child with a history of severe allergy to eggs.**

- A- Diphtheria.
- B- MMR.
- C- Tetanus.
- D- Yellow fever.
- E- Influenza.

### Answer Question 28

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

Patients with a history of anaphylaxis to eggs should not receive the egg-based vaccines: MMR, influenza and yellow fever.

NB. MMR vaccination should only take place where resuscitation facilities are available (e.g. in a hospital paediatric unit).

### Question 29

**In measles infection**

- A- The eruptive stage is infectious.
- B- Forchheimer spots are seen.
- C- The incubation period is 14-21 days.
- D- Koplik spots are pathognomonic.
- E- There are EEG abnormalities during infection in up to 5% of cases.

### Answer Question 29

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

Measles is infectious in the pre-eruptive stage, not the eruptive stage.

Forchheimer spots are palatal petechiae which are classically seen in Rubella.

The incubation period is short (7-14 days). Koplik spots are small grey lesions on the gum next to the 2<sup>nd</sup> molar and they are pathognomonic.

EEG abnormalities are seen in the acute disease in around 50% of cases.





### Question 30

#### Lyme disease

- A- Is a Rickettsial infection.
- B- Is transmitted by sandflies.
- C- May be diagnosed by measuring serum antibodies.
- D- Is a cause of erythema marginatum.
- E- Causes a chronic recurrent arthritis in most cases if untreated.

### Answer Question 30

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

Lyme disease is caused by the spirochaete, *Borellia burgdorfei*, and is transmitted by ixodid ticks on deer and sheep. The diagnosis is by measuring the IgM antibodies. The first feature is Erythema chronicum migrans (a painless red rash, spreading outwards). Most cases will develop a chronic arthritis within months to years without treatment.

### Question 31

#### In Rabies

- A- The virus enters at the bite wound and spreads via the lymphatics.
- B- There is a vaccine which is live attenuated.
- C- The average incubation period is less than 4 weeks.
- D- Aerophobia is pathognomonic.
- E- Dumb rabies is acquired from bats.

### Answer Question 31

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

Rabies enters via the bite, replicates in the muscle locally and spreads via the peripheral nerves to the brain where it replicates further.

The vaccine is a killed organism vaccine.

Rabies has an incubation period of 1-3 months on average, though it may be much longer.

Aerophobia is pathognomonic, but hydrophobia is seen in around half of cases.

Dumb rabies is a form involving a symmetrical ascending paralysis and is spread by bats.





### Question 32

**The following are true regarding the enteroviruses.**

- A- They may cause an acute haemorrhagic conjunctivitis.
- B- Coxsackie A and B are the most common causes of Ludwig's angina.
- C- They are the most common cause of aseptic meningitis.
- D- Rotavirus is one of the enteroviruses.
- E- The major cause of hand foot and mouth disease is Coxsackie A10.

### Answer Question 32

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

Acute haemorrhagic conjunctivitis may be caused by enteroviral infection; with enterovirus 70 identified in epidemics. Adenovirus classically causes epidemic conjunctivitis.

Ludwig's angina is a diffuse infection of the submandibular and sublingual spaces, and is usually bacterial.

Coxsackie A and B are the most common causes of Herpangina.

The enterovirus include Poliovirus, coxsackie viruses A and B, echoviruses and enteroviruses.

Coxsackie A16 is the major cause of hand and mouth disease.

### Question 33

**Urea cycle defects**

- A- Are associated with raised serum ammonia levels.
- B- Initial diagnosis is usually made by plasma amino acid profile and urine orotic acids.
- C- Include isovaleric acidemia.
- D- Are all Autosomal recessive disorders.
- E- May present with developmental delay.

### Answer Question 33

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

Urea cycle defects are a group of disorders involving defects of metabolism of ammonia in the urea cycle. They include carbamylphosphate synthetase deficiency (CPD), ornithine transcarbamylase deficiency (OTC), arginosuccinate synthetase (AS) deficiency, argininosuccinate lyase (AL) deficiency, arginase deficiency and N-acetylglutamate synthetase deficiency.

Initial investigations include a significantly raised serum ammonia level (usually >200 micro mol/l). plasma amino acid profile and urine orotic acid analysis usually make the initial diagnosis, which is confirmed by enzyme analysis.

Long-term management is with dietary protein restriction, avoidance of catabolic states and individual supplements.





### Question 34

**Chondrodysplasia punctata is seen in.**

- A- Contradi-Hunermann syndrome.
- B- Citrullinaemia.
- C- Zellweger syndrome.
- D- X-linked adrenoleucodystrophy.
- E- Warfarin toxicity.

### Answer Question 34

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

Chondrodysplasia punctata is a stippled appearance of the epiphysis of bones which is apparent on X-ray. It is seen in a number of conditions including:

- \*Zellweger syndrome (a peroxisomal disorder).
- \*Rhizomelic chondrodysplasia punctata.
- \*Warfarin toxicity.
- \*Conradi-Hunermann.

### Question 35

**First line investigations in a metabolic screen include**

- A- Serum ammonia.
- B- Urine amino acids.
- C- Plasma amino acids.
- D- Enzyme analysis from fibroblast culture.
- E- Blood gas.

### Answer Question 35

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

First line investigations in a metabolic screen include:

- \*serum urea and electrolytes, glucose, ketones, ammonia, liver function tests, clotting screen, lactate and amino acids.
  - \*Blood gase.
  - \*Urine: organic acids and ketones.
  - \*CSF: lactate and glycine
- Enzyme analysis is required for definitive diagnosis.





### Question 36

**Short stature may be caused by**

- A- Noonan syndrome.
- B- An XYY karyotype.
- C- Testicular feminisation syndrome.
- D- Chronic illness, if disproportionate.
- E- XY/XO mosaicism.

### Answer Question 36

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

Noonan syndrome and XY/XO mosaicism (turner syndrome mosaicism) both cause short stature.

XYY does not result in short stature, and neither does testicular feminisation syndrome (now less confusingly known as androgen insensitivity syndrome). Chronic illness results in proportionate short stature. Disproportionate short stature is seen in bone dyscrasias, for example, achondroplasia.

### Question 37

**In polycystic ovary syndrome.**

- A- There may be secondary amenorrhoea.
- B- There is early onset osteoporosis.
- C- There is a raised FSH:LH ratio.
- D- Prolactin is moderately raised.
- E- The oral contraceptive pill may be used in treatment.

### Answer Question 37

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

Polycystic ovary syndrome is a condition involving multiple small ovarian cysts and hormonal imbalance. The clinical features include secondary amenorrhea or menstrual irregularity, hirsutism, obesity, acne and infertility. Osteoporosis is not a feature. There is mild hyperprolactinaemia, and a raised LH level, with a raised LH:FSH level. Testosterone levels are moderately raised and may be normal. The oral contraceptive pill may help, and the anti-androgen cyproterone may be used.





### Question 38

**In type I diabetes mellitus.**

- A- First presentation in children is usually under the age of 4 years.
- B- Presentation is most common in winter.
- C- Most children have islet cell antibodies on first presentation.
- D- A sibling of a child with the disease has a 1 in 40 risk of developing it also.
- E- There is an association with H1ADR4

### Answer Question 38

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

Type 1 diabetes mellitus most commonly presents in older children, though can occur at any age. It most commonly presents in the spring and autumn. About 80% of children have islet cell antibodies on first presentation. The disease is associated with HLA B8, DR3 and DR4. Siblings have a 1 in 20 chance of developing the disease. The risk for a child developing the disease is increased if a parent has it, and the risk is greater if the father has it.

### Question 39

**Infants of diabetic mothers are at risk of the following.**

- A- Respiratory distress syndrome.
- B- Sacral agenesis.
- C- Hyperglycaemia.
- D- Polycythaemia.
- E- Hypoplastic left colon

### Answer Question 39

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

Infants of diabetic mothers have a 3 times increased risk of congenital malformations. In particular they may have macrosomia, Hypoplastic left colon, sacral agenesis and congenital heart disease. The neonate is at increased risk of respiratory distress syndrome, Polycythaemia and hypoglycaemia.







### Question 40

#### Regarding aniridia

- A- Cataract is often associated.
- B- Visual acuity is unaffected.
- C- It may be seen with chromosome 11p deletions.
- D- It is mostly sporadic.
- E- All patients should be screened with regular renal ultrasound scans.

### Answer Question 40

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

Aniridia may be inherited as autosomal dominant, with chromosome 2p or chromosome 11p deletions. It may be also inherited as Autosomal recessive, and about a third of cases are sporadic. Cataract is often associated and visual acuity is markedly reduced. There may be Nystagmus and macular hypoplasia. There is a strong association with Wilms tumour, both in the inherited chromosome 11p deletion and in the sporadic forms, and therefore screening with renal ultrasound scans is necessary.

### Question 41

#### Absence seizures.

- A- Usually last less than 30 seconds.
- B- Usually are preceded by an aura.
- C- If typical, may involve myotonic movements.
- D- May have a post-ictal state.
- E- A typical 1 per second spike and wave pattern is seen on the EEG.

### Answer Question 41

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

Absence seizures if typical last less than 30 seconds are not preceded by an aura and do not have a post-ictal state. Complex absence seizures may have a motor component such as myotonic movements.

A 3 per second spike and wave pattern is seen on the EEG.





### Question 42

**The following may cause acute ataxia.**

- A- Varicella infection.
- B- Phenytoin.
- C- Joubert disease.
- D- Cerebellar tumour.
- E- Abetalipoproteinaemia.

### Answer Question 42

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

Acute cerebellar ataxia may be caused by varicella infection as a post-infectious as a post-infectious event and phenytoin therapy where it is a sign of toxicity. A cerebellar tumour may result in acute ataxia if there is a bleed in to it. Though usually it is a cause of chronic ataxia. Joubert disease and Abetalipoproteinaemia are cause of chronic ataxia. The latter secondary to vitamin E deficiency.

### Question 43

**In myasthenia gravis.**

- A- Edrophonium is used in treatment.
- B- The external ocular muscles are unaffected.
- C- There is an association with HLA-B8, DR3.
- D- There are IgA antibodies to acetylcholine receptors.
- E- The baby of a mother with myasthenia gravis may develop a transient form of the disease.

### Answer Question 43

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

In myasthenia gravis, edrophonium is used to diagnose the condition, and result in a short-lived relief from the symptoms. The external ocular muscles are affected and there is diplopia and a partial ptosis. It is associated with HLA-B8, DR3. there are IgG antibodies to acetylcholine receptors. Infants may develop a transient form of the condition as the antibodies cross the placenta.





### Question 44

#### Legg-Calve-Perthes disease

- A- Involves aseptic necrosis of the tibial tubercle.
- B- Is 5 times more common in males than females.
- C- Is bilateral in around half of cases.
- D- Is generally managed conservatively in a child under 6 years.
- E- Is one of the osteochondritides

### Answer Question 44

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

Legg-calve-perthes disease is one of the osteochondritides. It involves aseptic necrosis of the femoral head. It is 5 times more common in males. It is bilateral in around a fifth of cases. Children under 6 years are generally managed conservatively, while those over 6 are managed with exercises and bed rest, abduction casts and osteotomy if necessary.

### Question 45

#### Concerning iron poisoning.

- A- Activated charcoal is of proven benefit.
- B- Clinical improvement at a few hours will result in a better outcome.
- C- Pyloric stenosis is a well recognised complication.
- D- Severe hepatic necrosis may occur within 24 hours.
- E- Desferrioxamine can be given orally

### Answer Question 45

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

Activated charcoal is not of proven benefit, but stomach washout can be helpful. Apparent early clinical recovery may lead to a false sense of security, as later degeneration can occur. Pyloric stenosis can occur 2-4 weeks later as a result of scarring from local irritation. Hepatic necrosis can occur but if it does it takes 2-4 days for the cytochrome enzymes to cease to function, and for cell death to ensue. Oral desferrioxamine should not be given as it may theoretically increase iron absorption and is expensive.





### Question 46

#### Regarding anticonvulsants.

- A- The anti-epileptic effect of intravenous diazepam lasts 60-90 minutes.
- B- Phenytoin appears to reduce cognitive function more than carbamazepine or sodium valporate.
- C- The estimated risk of congenital malformations in women receiving phenytoin during pregnancy is around 2%
- D- A transient leucopenia and thrombocytopenia occurs in up to 10% of patients on carbamazepine.
- E- Phenytoin is used for absence seizures.

### Answer Question 46

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

Intravenous diazepam is effective for only 20-30 minutes (i.e. enough time for phenytoin to be given). A recent study in the USA found that phenytoin appears to reduce cognitive function more than carbamazepine or sodium valporate. There is an estimated 10% risk of the foetal hydantoin syndrome (craniofacial and limb abnormalities) in infants of women on chronic phenytoin therapy. However, a severe agranulocytosis and aplastic anaemia is seen in 1 in 500,000. Absence seizures are managed with ethosuximide and sodium valporate.

### Question 47

#### Dopamine.

- A- Has a half-life of 2 minutes.
- B- Causes release of nor-adrenaline from nerve endings.
- C- Increases renal blood flow.
- D- Is inactivated by acidic solutions.
- E- Exerts inotropic action via  $\beta_1$  receptors.

### Answer Question 47

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

Dopamine has a short half-life and therefore an infusion is necessary. A plateau is reached by 5 half-lives (i.e. about 10 minutes). It increases renal blood flow via dopamine receptors on the renal arteries. Alkaline, not acidic, solutions such as bicarbonate will inactivate dopamine.





### Question 48

**The following concerning vincristine are true.**

- A- Bone marrow suppression occurs about 48 hours after treatment.
- B- A transient neuropathy is a recognised side-effect.
- C- Extravasation generally causes little problem.
- D- It is cell cycle specific for the M-phase.
- E- The dose needs increasing in biliary tract disease.

### Answer Question 48

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

Bone marrow suppression starts at around 7 days and peaks at 10-14 days, returning to normal within 21-28 days. A polyneuropathy is typically seen, although SIADH, autonomic dysfunction and cortical blindness are all reported. Vincristine is extremely toxic to tissues and extravasation requires urgent action. Pain, swelling and poor blood return should be looked for. Vincristine binds to the mitotic spindle during the M-phase causing its termination. Vincristine is metabolised via the liver and therefore the dose should be reduced in biliary tract disease.

### Question 49

**Waardenberg syndrome**

- A- Includes deafness in 80% of cases.
- B- Includes vitiligo.
- C- Type II is caused by mutations in the PAX3 gene.
- D- Includes mental retardation.
- E- Is associated with Hirschsprung's disease

### Answer Question 49

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

Waardenberg syndrome involves partial albinism, not vitiligo. The albinism usually involves a white forelock and pale blue or heterochromic irises. The hair become prematurely grey or white. Deafness is present in 25% of type I cases, and 50% of type II cases.

The disorder has been classified in to type I (including lateral displacement of the inner canthi) caused by mutations in the PAX3 gene on chromosome 2q35. and type II (no inner canthi displacement) caused by mutations in the human microphthalmia gene at chromosome 3p12.3-14.1. both types of Autosomal dominant inheritance.

Hirschsprung's disease may be associated along with oesophageal and anal atresia...





### *Question 50*

#### **Systemic-onset juvenile chronic arthritis.**

- A- Is a clinical diagnosis of exclusion.
- B- Classically involves a salmon-pink rash.
- C- Amyloidosis is a late feature.
- D- RhF is usually positive.
- E- A polyarthritis rarely occurs.

### *Answer Question 50*

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

Systemic-onset JCA is of equal sex incidence and may occur at any age of childhood. It is a clinical diagnosis, and the differential diagnosis include lymphoma, other malignancies, vasculitis, infection and other connective tissue diseases.

Clinical features are intermittent fever, a variable rash which is classically salmon-pink, however, myalgia, Arthralgia, hepatosplenomegaly, and lymphadenopathy. Pericardial effusions may occur. A polyarthritis occurs within a few months of disease onset in around half of cases. Late features do include Amyloidosis, as well as short stature and Micrognathia.

ANA and RhF are negative...





### Question 51

#### In juvenile onset dermatomyositis

- A- 'En coup de sabre' lesion on the forehead may be seen.
- B- There is a low mortality if untreated.
- C- It is associated with DQA1 \*0501.
- D- A heliotrope violaceous rash over the eyelids is pathognomonic.
- E- Subcutaneous calcium deposits may be seen.

### Answer Question 51

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

Juvenile onset dermatomyositis is a multisystem disease with inflammation of striated muscle and cutaneous lesions. The HLA association includes B8, DR3 and DQA1\*0501. If untreated, the mortality is high (up to 40%) but if treated, it is around 2-5%, though many (30-40%) will remain disabled. Clinical features include muscle pain with proximal muscle weakness and dysphagia, palatal regurgitation and respiratory muscle weakness. Cutaneous features include the heliotrope rash over the eyelids which is pathognomonic. Nail fold capillaries are also seen (look hard for these), and Gottron papules (red lumps over the DIP, PIP and knee joints). A butterfly rash over the face may be seen and subcutaneous calcium deposits may occur and these may extrude. Other features include joint involvement (Arthralgia, arthritis and contractures), gastrointestinal (ulcerations and bleeding), myocarditis, nephritis, CNS disease, interstitial lung disease, pulmonary haemorrhage, hepatosplenomegaly and retinitis.

### Question 52

#### Causes of blue sclerae include

- A- Ehler Danlos Syndrome.
- B- Osteogenesis imperfecta type IV.
- C- Marble bone disease.
- D- Pseudoxanthoma elasticum.
- E- Phenylketonuria.

### Answer Question 52

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

Blue sclera are seen in a number of diseases, involving defects in collagen. These include: Ehlers-Danlos syndrome, Marfan's syndrome, Pseudoxanthoma elasticum and Osteogenesis imperfecta types I, II and III (type III variable).

In phenylketonuria, blue irises are seen if the disease is untreated.







### *Question 53*

#### **The polymerase chain reaction.**

- A- Is a technique to sequence DNA.
- B- Can be used on very long DNA sequences (i.e. several kb long).
- C- May be used in antenatal chorionic villous sampling.
- D- Involves cycles of heating and cooling.
- E- May be used on DNA in a sample of blood stain that is several years old.

### *Answer Question 53*

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

The polymerase chain reaction (PCR) is a technique to rapidly make millions of copies of DNA which can then be used for analysis. It makes it possible to analyse DNA from very small sample of blood (i.e. a blood stain which may be several years old). PCR can be employed in antenatal chorionic villous sampling to enable rapid diagnosis (diagnosis in one day rather than one week). PCR can not amplify very long sequences, and thus can not be used to detect very large deletions.

The technique of PCR involves heating genomic DNA to denature it and produce single strands. Then the DNA is exposed to primer sequences which anneal to the complementary base pairs as the DNA is cooled to an annealing temperature. These primers flank the region of interest. The reaction is then heated to an intermediate temperature, and the primer sequence is extended as a base pairs are added by DNA polymerase. This double stranded DNA is heated again to denature it. The heating cooling cycle is repeated, and the newly formed DNA acts as a template for further DNA formation. Thus the number of copies doubles in each cycle by a chain reaction, and millions of copies of the original DNA are formed after several repeat cycles.





### *Question 54*

**Diseases associated with tri-nucleotide repeat expansions include.**

- A- Friedreich's ataxia.
- B- Miller-Dieker syndrome.
- C- Huntington's disease.
- D- Myotonic dystrophy.
- E- Myoclonic epilepsy with ragged red fibres (MERRF)

### *Answer Question 54*

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

Trinucleotide repeat expansion is seen in some inherited conditions, and successive generations may become more severely affected as the expansion occurs. The diseases associated with Trinucleotide repeat expansion include:

\*Huntington's disease, spinocerebellar ataxia type 1,2,3 and 6 (CAG repeat sequence, expansion occurs more often through the father) and myotonic dystrophy (CTG) and Fragile X syndrome (CGG) (expansion through the mother), and Friedreich's ataxia (GAA) repeat sequence, expansion through either parent.

\*Miller-Deiker is a microdeletion syndrome of chromosomal deletion 17p13.3.

\*MERRF is a mitochondrial DNA mutation disease.





### *Question 55*

**The following are true of Trisomy 21.**

- A- PDA is the commonest cardiac anomaly seen.
- B- Hypotonia is common in infancy.
- C- Risk of recurrence is 100% if a parent has the translocation 21:21.
- D- The genetic cause is non-disjunction in approximately 75% of cases.
- E- Karyotyping is essential

### *Answer Question 55*

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

Trisomy 21 (Down's syndrome) has an overall incidence of 1:650. The risk increases with maternal age, with an incidence of 1:100 to those of age 40 years. The genetic causes are non-disjunction (95%), Robertsonian translocation (4%) and mosaicism (1%).

Variable recurrence risk is seen, depending on the cause, and for this reason it is essential that chromosome studies are done.

For non-disjunction, the risk of recurrence is 1:200 if mother <35, and twice the age specific rate if mother >35 years.

In Robertsonian translocation the recurrence risk is 10-15% if the mother is the carrier, and 2.5% if the father is the carrier. The risk is 100% if a parent carries the translocation 21:21. the risk is >1% if neither parent carries a translocation.

Hypotonia is a consistent feature in infancy, occurring in approximately 80% of babies, and improving with age. Cardiac anomalies occur in 40% of infants. The commonest type is AVSD, then VSD, then PDA and then ASD.





### Question 56

**Regarding significance tests.**

- A-A paired t-test is used to compare two independent samples.
- B-The Mann-Whitney test is used on parametric data.
- C-The unpaired t-test is used on non-parametric data.
- D-Wilcoxon is the non-parametric equivalent of the paired t-test.
- E-The Chi-squared test may be used on parametric or non-parametric data.

### Answer Question 56

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

- \*The paired t-test is used to compare two dependant samples.
- \*The Mann-Whitney test is used on non-parametric data.
- \*The unpaired t-test is used on parametric data.
- \*Wilcoxon is the equivalent of the paired t-test used on non-parametric data.
- \*The Chi-squared test may be used on frequencies obtained from parametric or non-parametric

### Question 57

**In Erb's palsy.**

- A- The injury is to the C7,C8 and T1 nerve roots of the brachial plexus.
- B- The arm is classically held in the Waiter's tip position.
- C- There is a wrist drop.
- D- All will recover fully over weeks to months.
- E- Physiotherapy is necessary to prevent contractures.

### Answer Question 57

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

Erb's palsy results from damage to the upper nerve roots (C5 and C6) of the brachial plexus. The arm is held in abduction, with the elbow extended, the forearm pronated and the wrist flexed (the waiters tip position). A wrist drop occurs in Klumpke's palsy. Most , but not all will recover fully after several months. Physiotherapy is necessary, and prevents contractures developing.





### Question 58

The following are true of infants of maternal drug abusers.

- A- Rhinorrhea is a feature of drug withdrawal.
- B- Decreased sensory stimuli have a beneficial effect on the drug-withdrawing infant.
- C- Lacrimation is increased.
- D- The infants have an increased metabolic rate.
- E- There is an increased risk of SIDS.

### Answer Question 58

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

Infant of maternal drug abuses show many signs of drug withdrawal for which they need to be monitored and managed. Increased Lacrimation and rhinorrhea are features of drug withdrawal. These infants have an increased basal metabolic rate. They are managed with conservative measures which include decreased sensory stimuli, frequent feeds and swaddling. Drug treatment may be with opiates and sedatives as necessary. These infants are at increased risk of SIDS.

### Question 59

The following are causes of unconjugated neonatal hyperbilirubinemia.

- A- Biliary atresia.
- B- Gilbert's disease.
- C- Alagille syndrome.
- D- Hirschsprung's disease.
- E- Galactosaemia.

### Answer Question 59

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

Unconjugated neonatal hyperbilirubinaemia may be caused by Gilbert's syndrome, Hirschsprung's disease and galactosaemia. Biliary atresia and Alagille syndrome cause conjugated hyperbilirubinaemia.





### *Question 60*

#### **Regarding neonatal seizures.**

- A- They are usually terminated if the limb is held.
- B- If myoclonic, they often indicate a severe brain disturbance.
- C- Most neonatal seizures are obvious.
- D- There are usually EEG changes.
- E- They may respond to riboflavin.

### *Answer Question 60*

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

Neonatal seizures are often difficult to recognise, and may present simply as apnoea. One way of differentiating them from jitteriness is that they do not stop if the limb is held myoclonic seizures are concerning as they often indicate a severe underlying abnormality.

Most neonatal seizures are not obviously associated with EEG changes. A few seizures will be due to pyridoxine deficiency and respond to its replacement.

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# الكلمة الطيبة صدقة

