Hematology

Theme : Haematological Disorders

A. Aplastic anaemia
B. Beta Thalassaemia Major
C. Elliptocytosis
D. Glucose 6 Phosphate dehydrogenase deficiency
E. Haemochromatosis
F. Haemophilia A
G. Immune thrombocytopenia
H. Polycythaemia
I. Pyruvate Kinase deficiency
J. Sickle Cell anaemia

Select the most appropriate haematological diagnosis from the above list that best explains the following scenarios.

1) May be a consequence of parvovirus infection.	**A. Aplastic anaemia**
2) May present with a characteristic skin pigmentation and is secondary to multiple transfusions.	E. Haemochromatosis
3) Occurs as a result of a congenital defect of the red cell membrane.	**C. Elliptocytosis**

Comments:
Aplastic anaemia or bone marrow aplasia results in anaemia, neutropenia and thrombocytopenia. It may be congenital for example Fanconi's anaemia or acquired for example as a result of infection with for example Parvovirus infectio or due to drugs such as Chloramphenical, Sulphonamides or Chemotherapy. Haemochromatosis occurs when the structure or function of organs is deranged because of excessive Iron storage. Patients may exhibit bronzed skin and may develop Hepatitis or diabetes. Treatment requires chelation. Elliptocytosis is similar to spherocytosis in that the red blood cells exhibit a defect in the membrane. In Elliptocytosis the red blood cells are oval and the condition is benign. Haemolysis occurs with some forms of the disease.

Theme : Malignancy

A. Acute leukaemia
B. Ewing's Osteosarcoma
C. Lymphoma
D. Melanoma
E. Neuroblastoma
F. Retinoblastoma
G. Sarcoma
H. Thyroid cancer
I. Testicular seminoma
J. Wilm's tumour

Select the most appropriate chocie from thee above list for the following scenarios.

1) A condition which affects children under the age of 3, associated with a genetic locus on Chromosome 13.	F. Retinoblastoma
2) A tumour known that is recognised to spontaneously regress.	E. Neuroblastoma
3) A jaundiced baby on the postnatal ward is described as having blueberry muffin spots on the skin.	A. Acute leukaemia

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Comments:
Retinoblastoma is a tumour in the posterior part of the retina. It is associated with a deletion of the long arm of chromosome 13. The average age of diagnosis is 8 months for bilateral involvement ands 24 months for unilateral involvement. It may present with leucocoria, visual loss or a squint. A Neuroblastoma may arise anywhere where neural crest cells migrate. The median age of diagnosis is approximately 2. In babies less then 1 year of age or in the early stages of tumour development spontaneous regression has been known to occur. Acute lymphoblastic leukaemia is the commonest malignancy of childhood. Initial symptoms may be non-specific for example lethargy and irritability. Ultimately there is bone marrow failure which presents with pallor, thrombocytopenia and neutropenia. Diagnosis is made by seeing blasts on a peripheral smear There may be anaemia, thrombocytopenia and the white cell count may be raised or low. Babies born with congenital leukaemia have lesions over the skin which are described as being similar to blueberry muffins spots

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1) May be exacerbated by exposure to anti-malarial therapy	**D. Glucose 6 Phosphate dehydrogenase deficiency**
2) Characterised by the overgrowth of bones of the face.	**B. Beta Thalassaemia Major**
3) May be an associated feature of congenital cyanotic heart disease.	H. Polycythaemia

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Comments:
Glucose-6-phosphate dehydrogenase deficiency is an episodic haemolytic anaemia which can be exacerbated by certain drugs, in particular anti-malarial therapy. There is a racial difference in prevalence. Laboratory findings include low levels of G6PD and Heinz bodies present in the red blood cells. Thalassaemia is a group of hypochromic anaemia's due to abnormalities of globin chain synthesis. Regular blood transfusions are necessary and clinical features include compensatory hypertrophy of erythropoietic tissue in medullary and extra medullary spaces. For example liver, spleen and the marrow in the face which results in characteristic facies. Polycythaemia exists when red blood cell count, Haemoglobin and haematocrit exceed the upper limits of normal and may co-exist with chronic oxygen desaturation states, for example cyanotic cardiovascular and pulmonary disease.

Theme : Bleeding disorders

A. Antithrombin III deficiency
B. Congenital afibrinogenaemia

C. Haemophilia A
D. Haemophilia B
E. Haemophilia C
F. Idiopathic thrombocytopenic purpura
G. Protein C deficiency
H. Protein S deficiency
I. Vitamin K deficiency
J. Von Willebrands disease

Choose the most appropriate diagnosis from the above list that best describes the following coagulation abnormalities.

1) An autosomal dominant disorder, characterised by thrombotic events. — A. Antithrombin III deficiency

Note:
Antithrombin 3 deficiency prolonged in occlusion of blood vessels with platelet plugs resulting in thrombotic events.

2) Prolonged partial thromboplastin time, normal Prothrombin time. factor 8 deficiency — C. Haemophilia A

Note:
Haemophilia A results due to low factor 8 levels. The severity of the illness depends on the level of activity and patients often present with haemarthrosis.

3) An autosomal dominant disorder associated with a prolonged bleeding time. — J. Von Willebrands disease

Note:
describes Von Willebrand's disease an autosomal dominant condition which results in prolonged bleeding due to low Von Willebrand factor, which is needed for platelet aggregation. There is a prolonged bleeding time and treatment with FFP is sometimes required.

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Choose the most appropriate diagnosis from the above list that best describes the following coagulation abnormalities.

1) Normal Prothrombin time.
Elevated thromboplastin time, factor 9 deficiency. D. Haemophilia B

Note:
Haemophilia B also known as Christmas disease is an X linked recessive disorder. It is as a result of low Factor 9 levels.

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2) Prolonged Prothrombin time and partial thromboplastin time, normal bleeding time. I. Vitamin K deficiency

Note:
Item 2 describes Vitamin K deficiency which is necessary for carboxylation of factors 2, 7 ,9 and 10.

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3) Bruising and petechiae over lower extremities with low platelets. **F. Idiopathic thrombocytopenic purpura**

Note:
fits the diagnosis of idiopathic thrombocytopenic purpura. A condition associated with mucocutaneous bleeding and petechiae often secondary to viral infections and the platelet count is less than 20 x 10/9. Treatment options include steroids and gammaglobulin.

Theme : Malignancy

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C. Lymphoma
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E. Neuroblastoma

F. Retinoblastoma
G. Sarcoma
H. Thyroid cancer
I. Testicular seminoma
J. Wilm's tumour

Select the most appropriate choice from the above list for the following scenarios

1) A 3 year old with pallor, bruising and hepatosplenomegaly.	**A. Acute leukaemia**
2) A 16 year old with shortness of breath and night sweats and a cervical lymphnode.	C. Lymphoma
3) A 4 year old known to have a deletion of Chromosome 11 presents with an abdominal mass.	**J. Wilm's tumour**

Comments:
Acute lymphoblastic leukaemia is the commonest malignancy of childhood. Initial symptoms may be non-specific for example lethargy and irritability. Ultimately there is bone marrow failure which presents with pallor, thrombocytopenia and neutropenia. Diagnosis is made by seeing blasts on a peripheral smear There may be anaemia, thrombocytopenia and the white cell count may be raised or low. Babies born with congenital leukaemia have lesions over the skin which are described as being similar to blueberry muffins spots. Lymphoma is divided in to Hodgkin's and Non-Hodgkin's lymphoma. It arises in lymph nodes and enlarged nodes may be firm and non-tender. Mediastinal involvement may cause a chronic cough or bronchial / tracheal compression. Symptoms include night sweats, persistent fever and weight loss . Wilm's tumour is a solitary growth in a part of the kidney. It is associated with deletions of chromosome 11 (the probable location of the tumour suppression gene). It may present with an abdominal mass or haematuria. It is also associated with genital urinary anomalies, aniridia, hemihypertrophy and Beckwith-Wiedemann syndrome.

Theme : Diagnosis of neonatal jaundice.

A. ABO incompatibilty
B. Breast milk jaundice
C. Cephalhematoma
D. Drug allergy
E. Extrahepatic biliary atresia
F. Galactosaemia
G. G6PD Deficiency
H. Neonatal sepsis
I. Physiological jaundice
J. RH Incompatibility

For each of these jaundiced babies below choose from the list above the single most likely diagnosis. Each option may be chosen more than once or not at all.

1) A mother has blood group A Rhesus positive. Her baby is blood group B Rhesus negative.

A. ABO incompatibilty

Note:
This mother would produce antibodies to the baby's B blood group and hence haemolysis with jaundice occurs.

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2) A neonate is severely jaundiced with reducing substances noted on urine dipstick.

F. Galactosaemia

Note:
A typical picture of **galactossaemia**. In the newborn period, infants present with an acute encephalopathy. In untreated patients, there is severe liver disease, mental retardation, epilepsy and choreoathetosis.

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3) A 4 day old baby is well but has a tinge of jaundice.

I. Physiological jaundice

Note:
Common in the first week of life.

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4) A breast fed 3 week old baby has mild jaundice but is gaining weight satisfactorily.

B. Breast milk jaundice

Note:
Common from the 2nd to 5th days of life.

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5) A 12 day old baby is jaundiced with pale stools.

E. Extrahepatic biliary atresia

Note:
Biliary atresia. is a progressive inflammatory process that begins very soon after birth. On average, there is one case of biliary atresia out of every 15,000 live births. Females are affected slightly more often than males. In the United States, approximately 300 new cases are diagnosed each year.