Hemoglobinopathies – Part 2

COMPLICATION :

1- Growth and sexual retardation .
2- Gallstones (pigmented) .
3- Splenic dysfunction .... Infecction with pneumococci and encapsulated organisms like H. infleunza
4- Renal papillary necrosis and renal failure .
5- Bone complications :

a.Aseptic necrosis of the femoral or humeral heads.
b.Salmonella osteomyelitis .
c.The hand – foot syndrome which is caused by painful infarcts of the digits .

6- Iron overload due to repeated blood transfusions
7- Folate deficiency .
8- Chronic leg ulcers .

Investigation

1- normochromic Normocytic anaemia .
2- Increased reticulocyte count .
3- Increased serum indirect bilirubin .
4- Blood film :
   a. sickle cells .
   b. nucleated RBCs .
   c. target cells .
5- Hb electrophoresis : show
....>90 % Hb S .
....No Hb A .
....2-10 % Hb F .
6- Skull X-R : Hair on End appearance .

Causes of death

1- Infection .
2- Organ infarction .
3- Heart failure and respiratory failure .
4- Acute chest syndrome .
5- Iron overload .

TREATMENT :

I- During steady state :

1- good nutrition .
2- folate supply .
3- blood transfusion in decreasing the Hb .
4- penicillin V to protect against pneumococcal infection which may be lethal for patients with hyposplenism .
5- Vaccination against pneumococcus and if available H.influenza and HBV .

II- During Crisis :

1- Aggressive rehydration .
2- O_2 therapy .
3- Adequate analgesia (morphine 0.1-0.15 mg/ kg every 3-4 hrs) to control the severe pain . Inhalation of nitrous oxide can provide short-term pain relief .
4- Antibiotics .
5- A regular transfusion programme to suppress Hb S production and maintain the Hb S < 30 % may be indicated in reccurent severe complications such as CVA in children or chest syndromes in Adult .

6- Exchange transfusion , where a patient is Simultaneously venesected and transfused to replace HbS with HbA , may be used in the following indications :

a. Life-threatening crises .
b. To prepare patients for surgery .
c. Acute chest syndrome .
d. Stroke .
e. Bone marrow necrosis .
f. Priapism .

The goal of exchange transfusion is to achieve a level of 30-40 % HbS .

Hydroxyurea

It is most significant advance in the therapy of SCA.
It's indicated in :

1- patients with severe symptoms .
2- patients experiencing repeated episodes of acute chest syndrome .
3- with > 3 crises / year .

Hydroxyurea (10-30 mg/kg / day) increases fetal Hb and may exert beneficial affects on red cell hydration , vascular wall adherence and suppression of the granulocyte and reticulocyte counts .

White cells and reticulocytes may play a major role in the pathogenesis of SC crisis , and their suppression may be an important benefit of hydroxyurea therapy .

BONE MARROW TRANSPLANTATION
It can provide definitive cures but is known to be effective and safe only in children .

GENE THERAPY for SCA is being pursued, but no safe measures are currently available .

** Agents blocking RBC dehydration or vascular adhesion , such as **clotrimazole** or **magnesium** , may have value as an adjunct to hydroxyurea therapy (these 2 drugs are being evaluated in clinical trials .

Thank You,,,

1-True or false
The following produce hemolysis in patients with G6PD deficiency :
A primaquine
B Penicillin
C Tetracycline
D Glandular fever
E Nitrofurantoin

- The following is not true with refersnce to Hereditary spherocytosis :
A Inheritance is autosomal dominant .
B May be complicated by megaloblastic crises .
C Leg ulceration is a feature .
D Osmotic fragility is reduced .
E Splenectomy is the treatment of choice .