
Ihsan Mardan Humood

Name :Ahmed Bassam Idrees

Date of birth: 1st/Sept./2004

Sex : Male

Occupation: Student at primary school, 3rd class

Chief complaint: chronic case of anemia since 1 year old.

History of present illness: the mother noticed pallor and poor feeding of her child after the first year of his age, they consulted a doctor who requested CBP.

18/1/2006	1.4 years old	N.R. 2-6 Y
Hb	: 4.0 g/dl	11-14
PCV	: 9 %	34-40
WBC	: 7 × 10^9/L	5-15
Neutrophils	: 35 %	
Lymphocytes	: 55 %	
Monocytes	: 10 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: Adequate	
Reticulocytes	: 0.1 %	

RBC : Normochromic oval macrocytic.
WBC : Mature cells.
CONCLUSION: Severe MACROCYTIC anemia.

The patient was admitted to hospital and received packed red blood cells, then discharged with tonics.

Till the age of two years old, he received 4 times packed red blood cells without improvement of his condition apart from short period after blood transfusion. So his doctor advised the family to do bone marrow aspiration.

3/10/2006	2 years old	N.R. 2-6 Y
Hb	: 6.0 g/dl	11-14
PCV	: 18 %	34-40
WBC	: 4 × 10^9/L	5-15
Neutrophils	: 50 %	
Lymphocytes	: 47 %	
Monocytes	: 3 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: Adequate	
Reticulocytes	: Zero %	

RBC : Normochromic normocytic **WBC**: Mature cells

CONCLUSION: Severe Normochromic anemia with zero reticulocyte count

Bone marrow aspiration report 3/10/2006

Cellularity : Normocellular marrow.

Megakaryocytes: Adequate

Erythropoiesis : Absent

Leucopiesis : No increase in immature cells
Active granulopoiesis
Lymphocytes seen in increased number.
No abnormal cell infiltrate.

Conclusion: Absent erythroid activity.

Final diagnosis: PURE RED CELL APLASIA.

Prednisolon tab. 25mg/day (5mg×5) was added to the patient. His condition was improved over time.

16/11/2006		N.R. 2-6 Y
Hb	: 13.0 g/dl	11-14
PCV	: 39 %	34-40
WBC	: 4 × 10^9/L	5-15
Neutrophils	: 55 %	
Lymphocytes	: 42 %	
Monocytes	: 3 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: Adequate	
Reticulocytes	: 5 %	

RBC : Normochromic macrocytic.
WBC : Mature cells.
PLT : Adequate.

Then tapering of steroid was gradually with follow up of Hb and reticulocyte.

16/12/2006		N.R. 2-6 Y
Hb	: 13.8 g/dl	11-14
PCV	: 42 %	34-40
WBC	: 6 × 10^9/L	5-15
Neutrophils	: 68 %	
Lymphocytes	: 30 %	
Monocytes	: 2 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: Adequate	
Reticulocytes	: 0.1 %	

RBC : Normochromic normocytic/macrocytic
WBC : Mature cells.
PLT : Adequate.

The doctor advised the family according to the reticulocyte count to continue on 10mg/day of prednisolon tab.

14/9/2009		N.R. 2-6 Y
Hb	: 12.5 g/dl	11-14
PCV	: 38 %	34-40
WBC	: 4 × 10^9/L	5-15
Neutrophils	: 70 %	
Lymphocytes	: 27 %	
Monocytes	: 03 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: Adequate	
Reticulocytes	: 2 %	

RBC : Normochromic normocytic/macrocytic
WBC : Mature cells.
PLT : Adequate.

According to the last CBP on September 2009, the doctor decided to taper the steroid gradually.

13/4/2011	7 years old	N.R. 6-12 Y
Hb	: 11.0 g/dl	11.5-15.5
PCV	: 34 %	35-45
WBC	: 6 × 10^9/L	5-13
Neutrophils	: 55 %	
Lymphocytes	: 44 %	
Monocytes	: 1 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: Adequate	
Reticulocytes	: Zero %	

RBC : Normochromic normocytic/macrocytic
WBC : Mature cells.
PLT : Adequate.

The patient returned to prednisolon tab. 10mg/day, and he continued till now on 2.5mg every other day.

History of other systems: nothing of significant.

Family history: he is the fourth child of non relative parents, nobody has the same condition.

Drug history: no allergy to drugs, Hx of prednisolon only.

On examination:

9 years old child, looks well, no pallor, no jaundice, no LAP

No acne, both height (132cm) and weight (30kg) are normal for his age, no other signs of steroid complication.

Chest: clear, NVB.

Abdomen: soft, no organomegaly.

- **US of the abdomen:** 2011

Normal liver, both in size and texture.
Normal spleen.
Normal both kidneys.

- **Echocardiogram** 2011

was normal.

- **CT and MRI examination of the chest:** 2011

The thymus gland is enlarged and of well defined smooth margin and has no focal abnormal enhancement.

Comment: No evidence of thymoma with mild hepatomegally and abnormal splenic texture.

13/10/2013	9 years old	N.R. 6-12 Y
Hb	: 11.7 g/dl	11.5-15.5
PCV	: 35 %	35-45
WBC	: 7 $\times 10^9$/L	5-13
Neutrophils	: 59 %	
Lymphocytes	: 40 %	
Monocytes	: 1 %	
Eosinophils	: 0 %	
Basophils	: 0 %	
Platelet in film	: 290 $\times 10^9$/L	
Reticulocytes	: 0.6 %	

RBC : Normochromic normocytic/macrocytic
WBC : Mature cells.
PLT : Adequate in blood film.

TABLE 17-4. RED CELL APLASIA: CAUSES

Congenital
Diamond–Blackfan syndrome
Acquired
Primary
Autoimmune: immunoglobulin inhibitors of erythroid precursors or of erythropoietin
T-cell inhibition of erythroid precursors
transient erythroblastopenia of childhood
Secondary
Tumors: thymoma Lymphoma: Hodgkin non-Hodgkin chronic lymphocytic leukemia large granular lymphocytic leukemia acute lymphoblastic leukemia other tumors
Infections: parvovirus (transient) human immunodeficiency virus viral hepatitis infectious mononucleosis others
Immune disorders: systemic lupus erythematosus rheumatoid arthritis
Drugs and chemicals (e.g., benzene, diphenylhydantoin, isoniazid)
Nutritional deficiencies: riboflavin vitamin B_{12} or folate deficiency

Inherited pure red cell aplasia

Diamond-Blackfan anaemia (DBA)

It is a haemopoietic stem cell disorder of which the earliest manifestation is pure red cell aplasia. Later neutropenia and thrombocytopenia may also develop.

Inheritance is usually autosomal dominant but in some families is autosomal recessive.

Inherited pure red cell aplasia

About three-quarters of cases appear to be sporadic.

About 40% of patients have associated congenital abnormalities such as craniofacial, thumb, cardiac and urogenital malformations. The incidence of AML is increased.

The diagnostic criteria for DBA have comprised:

(i) Normochromic, usually macrocytic, but occasionally normocytic anaemia developing in early childhood.
(ii) Reticulocytopenia.
(iii) Normocellular bone marrow with selective deficiency of erythroid precursors (erythroblasts < 5%).
(iv) Normal or slightly decreased leucocyte counts.
(v) Normal or often increased platelet counts.

Inherited pure red cell aplasia

More recently, elevated erythrocyte deaminase activity, macrocytosis and elevated fetal haemoglobin have been added to the list of supportive features of DBA.

It has also been recognized that in a subset of cases the presentation may be in adulthood.

Inherited pure red cell aplasia

Further tests:

Serum soluble transferrin receptor is greatly reduced in all types of pure red cell aplasia.

Inherited pure red cell aplasia

The first line of treatment for DBA remains corticosteroids. Once a maximal haemoglobin response has been achieved, the dose of prednisolone should be tapered slowly until the patient is on the lowest dose possible on an alternate - day regimen.

Acquired pure red cell aplasia

Acquired pure red cell aplasia may be transient or persistent.

Transient pure red cell aplasia is often caused by parvovirus B19 infection and, unless the patient has, coincidentally, a shortened red cell lifespan, is so brief that it often goes undiagnosed.

Acquired pure red cell aplasia

Chronic pure red cell aplasia may be immunological in origin, as when it is associated with thymoma, autoimmune disease, large granular lymphocyte leukaemia or chronic lymphocytic leukaemia.

Acquired pure red cell aplasia

Acquired pure red cell aplasia is associated with a thymoma in approximately 50% of patients, although it complicates only approximately 5% of thymomas.

Acquired pure red cell aplasia

Antibodies to erythroid precursors have been demonstrated in some patients, and removal of the thymoma (which is usually benign) leads to resolution of the anaemia in about half of those affected.

Acquired pure red cell aplasia

Immunosuppressive therapy with cyclophosphamide, ciclosporin, steroids or plasma exchange may be helpful in patients who relapse.

There is no evidence that erythropoietin is useful in pure red cell aplasia.

Distinction between inherited and acquired red cell aplasia may be impossible in the younger patient. william H.


Thank you