

Familial Erythrocytosis

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Abstract

Erythrocytosis is a rare red cell disorder characterized by an elevated haematocrit (Hct) and haemoglobin (Hb) level and red blood cells (RBC) with normal white cells and platelets. Both sporadic and familial forms exist. A 49 years old gentleman presented with lightheadedness, tinnitus, dizziness and uncontrolled hypertension of 5 days duration KIST Medical College Hospital Lalitpur Nepal. His Hb was 17.0 g/dl, Hct 69%, red blood cell (RBC) count 5.8 Million, white blood cell (WBC) 10500 and platelets 255000. His oxygen saturation was 98% and serum erythropoietin level was 11.3 m IU/ml (normal range 3.7-29.7 m IU/ml). Hb and Hct of his elder brother was 16.5 g/dl and 54% respectively and of elder sister 14.5 g/dl and 50% respectively. Our patient was having erythrocytosis most likely familial. Family didn't agree for genetic workup; this is the limitation of our case report.

Key words: Erythrocytosis, Familial, Nepal

Introduction

Erythrocytosis is a rare heterogeneous red cell disorder that can arise from diverse molecular origins. It is characterized by an elevated haematocrit (Hct) and haemoglobin (Hb) level and red blood cells (RBC). There is no accompanying increase in the number of white blood cells (WBC) and platelets. Both sporadic and familial forms exist and the presentation is highly variable (1, 2, 3, 4, 5, 6, 7). Many of the affected individuals are not aware of any abnormality, nor do they have any sense of illness as these individuals have sub-clinical condition (6). Some of the affected individuals have a serious condition presenting with cardiovascular events, and thrombo-embolic phenomena, and abnormal bleeding (2). Here we present a case of 49 years old gentleman with erythrocytosis.

Case Report

A 49 years gentleman, teacher and manager by profession presented in out patient department of KIST Medical College Hospital with lightheadedness, tinnitus and

dizziness of 5 days duration. He was found to be hypertensive at the age of 36 years, since then he has been taking tablet enalapril 5mg daily. With enalapril his blood pressure remained controlled till a few days back. Because of the mentioned symptoms and raised blood pressure he took tab atenolol 50mg daily in addition to enalapril for the last three days. He was operated for appendicitis in 1981. He used to donate blood very frequently in the past. His father was hypertensive and elder brother is diabetic and hypertensive.

At the time of presentation he looked to be tired. His weight was 83Kg and height 180cm. His pulse was 76 per minute regular, respiratory rate 16 breaths per minute, temperature 98 degree Fahrenheit and blood pressure 140/90 mm Hg. Except for appendectomy scar over abdomen, the physical examination was unremarkable.

His blood sugar fasting was 5.8 m mol/lit, urea 4.1 m mol/lit, creatinine 80 micro mol/lit, uric acid 327 micro mol/lit, cholesterol 4.3 m mol/lit, HDL 1.2 m mol/lit, LDL 2.0 m mol/lit, triglyceride 2.2 m mol/lit, LDH 568 IU/lit, SGOT

25 IU/lit and SGPT 16 IU/lit.

His Hb was 17.0 g/dl, Hct 69%, red blood cell count 5.8 Million, Morphology of red blood cells in peripheral blood smear was normal. WBC count was 10500 (neutrophils 67%, lymphocytes 29%, monocytes 3% and eosinophils 1%) and platelets 255000. His oxygen saturation with pulse oximeter was 98%. His serum erythropoietin level was 11.3 m IU/ml (normal range 3.7-29.7m IU/ml). His ECG and ECHO findings were within normal limit while USG abdomen revealed fatty changes in liver. With readjustment of dosages anti hypertensive medication (enalapril 5mg and atenolol 100mg daily) his blood pressure was controlled. He donated blood also.

With counseling his brother and sisters agreed for Hb and Hct tests. Hb and Hct of his elder brother aged 56 years was found to be 16.5 g/dl and 54% respectively, Hb and Hct of his elder sister aged 53 years 14.5 g/dl and 50% respectively and Hb of his younger sister aged 40 years was 15.5 g/dl.

Discussion

Our patient was having erythrocytosis as his Hb, Hct and RBC count was raised but WBC and platelets counts were normal; it was most likely familial as Hb and Hct of two other family members were found to be raised. (1, 2, 3, 4, 5, 6)

A large Finnish family with autosomal dominant erythrocytosis has been reported in the literature. The erythrocytosis had not had any obvious effect on the health or life span of the affected individuals, some of them had hypertension. In fact, many reached an advanced age and one (best cross-country skiers in the world) won several Olympic gold medals and world championships in sports treated successfully with repeated venous phlebotomies. (2, 6)

Melanie J. Percy et al (2008) reported a case of erythrocytosis in 89-year-old lady who was diagnosed at the age of 54 years. She had hypertension and diverticulitis. She used to have venesection two to three times a year till the age of 81 years; since then her Hb level has been between 14.0 and 15.5 g per deciliter, and Hct between 45% and 52%. (7)

Long standing hypertension of our patient could be due to erythrocytosis. Due to frequent blood donation in the past he might had not developed any other symptoms before.

His serum erythropoietin level was normal. Other authors also reported normal erythropoietin level in some of their cases. (1, 6)

For genetic work up family did not agree; so it could not be done. This is limitation of our case report.

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