

Benign familial polycythaemia in a young male

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Abstract

Polycythaemia has been reported rarely as a familial condition. There is evidence to suggest transmission as a Mendelian dominant trait, but recessive inheritance has also been described. We present here a case of benign familial polycythaemia in a 25-year-old male with similar presentation in his family members. Our patient presented with reddish discolouration of the eyes, early satiety, weight loss and itching at intervals, for four years. An additional examination revealed red beefy tongue and Grade III clubbing. The importance of presenting this case lies in the fact that the prognosis appears to be good in these patients, but regular observation is necessary as Kiladjan and colleagues have mentioned that there is a risk of leukaemia, thrombosis and myelofibrosis in these patients later on, as the idiopathic erythrocytosis group contains a certain number of polycythaemia patients.

Introduction

The term familial and congenital polycythaemia encompasses a heterogeneous group of disorders with the common characteristic of an absolute increased red cell mass since birth and/or similar phenotype is also present in relatives.¹ It presents at a younger age than classical polycythaemia, often in childhood. There may be few if any symptoms. Leukocytosis and thrombocytosis are absent. The prognosis appears to be relatively good and for this reason the condition is sometimes called *benign familial erythrocytosis*.

This is a case of benign familial polycythaemia in a young male (25 years old). There have been reports of similar cases. Of 25 patients reported in one series, 12 were found to have elevated erythropoietin levels and were therefore assumed to represent patients with secondary polycythaemia, these patients tended to be younger than the patients with normal erythropoietin levels.²

Case Report

A 25-year-old Muslim male patient, farmer by occupation, residing at Maldah district in West Bengal (India) presented with generalised weakness along with reddening of the eyes for the last four years with history of early satiety and weight loss. Patient had history of itching at regular intervals with clubbing also for the last four years.

Patient also gives similar history in his father (60 years old) and his sister (15 years old); his 11-year-old brother and his other sister (20 years old) were however not affected. His father and sister were asymptomatic.

There was no history suggestive of fever, drenching night sweats, lymphadenopathy, bleeding from any site of the body, history of loose motions, history of exposure, blood transfusion, dizziness, any visual, cardiologic and neurological symptoms. There was also no history of headache, vomiting, convulsions. Patient had no complaints regarding affection of renal system and he is non diabetic, non hypertensive.

On examination the patient had reddish discolouration of the lower palpebral conjunctiva, and beefy red tongue. There was Grade III clubbing without any rise of Jugular venous pulse (JVP) or oedema. Blood pressure, Pulse, Respiratory rate were within normal limits. There was two finger firm hepatomegaly and spleen was moderately enlarged, firm, nontender. There were no petechial spots, sternal tenderness or lymphadenopathy. O₂ saturation (SpO₂) was also normal, so were examination of the testes and per rectal examination.

Blood investigations revealed normal glucose, urea and creatinine levels. *Haemoglobin (Hb) was 25.3 gm/dL* while Total Leucocyte Count (T.L.C) was 5,800/mm³ [Neutrophil - 60%, Lymphocyte - 30%, Eosinophil - 6%, Monocyte - 4%], Platelets - 3,50,000/mm³ and Erythropoietin Sedimentation Rate (E.S.R) was 5mm (1st hr). Erythropoietin (EPO) levels were increased - 144 mU/mL, normal values ranging from 3.7 to 31.5 mU/mL. Hb electrophoresis, bone marrow examination revealed no abnormality. Philadelphia chromosome t(9;22) was not detected by Fluorescent In situ Hybridisation (FISH) technique. Other genetic mutations like JAK 2, V617F and exon 12 could not be studied as there was no facility to perform these investigations in our setup. Similarly Electrocardiography (ECG) and Echocardiography were unremarkable. Malarial slide and antigen were negative and so were Aldehyde test and RK 39 test for kalaazar. Chest X-ray (PA view) did not reveal any abnormality. Malignancies like renal cell carcinoma, hepatocellular carcinoma and adrenal adenoma were ruled out by a normal Ultrasonography

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(USG) of whole abdomen and Computed Tomography (CT scan) of abdomen while a normal CT scan of Brain ruled out Central Nervous System (CNS) hemangiomas. Twenty-four hour urinary catecholamines and metanephrines as well as blood levels of catecholamines and metanephrines were normal eliminating pheochromocytoma.

Father was asymptomatic. Laboratory parameters were remarkable for High Hb levels (25 g/dL) and raised EPO levels (140 µm/mL) with normal T.L.C and platelet count. Hb electrophoresis and bone marrow study were unremarkable. As in his son Philadelphia chromosome was not found. He did not come for follow up. The sister did not give consent for examination or investigation neither did she come for follow up.

Discussion

The term idiopathic polycythaemia (or erythrocytosis) refers to patients who have an unknown aetiology of polycythaemia even after extensive investigation. It could include most of the patient categorised as *benign erythrocytosis*. The existence of this group which is estimated to contain 20-30% of patients evaluated for polycythaemia³ essentially represents a failure to categorise all polycythaemia patients.

Our case is an interesting case of benign familial polycythaemia in whom detailed evaluation and investigation failed to detect any cause of polycythaemia in the patient as well as his father.

The prognosis of these patients appears to be relatively good. Some of these families have been identified as possessing a high O₂ affinity Hb variant, in others low erythrocyte 2.3 –

Biphosphoglycerate (2,3 BPG) has been found and in the third variety with recessive inheritance inappropriately increased erythropoietin secretion has been identified.⁴ Our patient typically represents the last variety mentioned with elevated erythropoietin levels. In a polycythaemic patient, establishing a correct diagnosis of a high affinity Hb variant is important as these patients have normal life expectancy and do not require phlebotomy.⁵ However in this case our 25 years old patient as well as his 60-year-old father had normal O₂ affinity Hb variant as indicated by the normal Hb electrophoresis. They had not required phlebotomy until now and whether it alters their life expectancy remains to be seen.

Conclusion

Although the prognosis is usually good in these patients, study done by Kiladjian *et al.* proved otherwise. They treated 39 patients

with idiopathic erythrocytosis with pipbroman and compared their clinical course to 140 concurrently treated polycythaemia patients.⁶ The risk of thrombosis, leukaemia and myelofibrosis was the same in the two groups.

Because this category probably represents a mixed bag, including early polycythaemia, mild secondary polycythaemia and normal individuals at the higher end of the bell shaped curve for red cell mass,⁷ a cautious approach is warranted. Observation may be the most reasonable intervention; this may be the patient subset in which otherwise low yield studies like erythropoietin levels and erythroid progenitor studies are likely to be useful.

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