# A Rare Case of Congenital Erythrocytosis

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**Background**

Erythrocytosis (increased RBC mass or polycythemia) maybe primary due to an intrinsic defect in the bone marrow stem cells/erythroid progenitor cells or secondary where the red cell production is driven by factors external to the erythroid compartment, such as increased erythropoietin production for any reason. Primary and secondary causes can be further classified into congenital and acquired causes. The classical acquired primary erythrocytosis is Polycythemia Vera where patients have an abnormal clone and frequently an increased white blood cell and platelet count. Majority of patients have a clone of cells with gain of function mutations in JAK2. Congenital causes include mutations of the erythropoietin receptor and defects in oxygen sensing pathways including VHL, PHD2 AND HIF2A mutations.

**Case**

15 yrs male presented with complaints of head ache, leg pain and redness in the eye with significant history of redness of the eye and weakness since birth. Clinical examination revealed splenomegaly. Laboratory investigations showed erythrocytosis with RBC count of 10.46 million/cumm, haemoglobin 22.7gm/dl, haematocrit 76.2%, normal WBC and platelet count. Lactate dehydrogenase levels was increased 925IU/L, renal function and liver function tests were normal. Bone marrow examination with FISH for t(9,22) and JAK 2 mutational analysis was negative which ruled out Polycythemia Vera. Serum erythropoietin levels was increased 290mIU/ml. Oxygen saturation studies was normal. Haemoglobin electrophoresis showed normal adult pattern. Ultrasound, CT scan and PET scan revealed no significant lesion known to cause secondary erythrocytosis. Diagnosis of congenital erythrocytosis ?cause was made. Since there was no provision locally for testing defects in oxygen sensing pathways it could not be done. The patient was managed with venesection and aspirin.

**Results**

A raised red cell count will increase blood viscosity and thus will have clinical consequences resulting in thromboembolic events. Once an erythrocytosis is established, identification of the cause is the next focus. It starts with a complete history and examination with exploration for secondary causes. EPO levels is an initial way to guide further investigations.The above case also emphasises the importance of EPO. There is little evidence to guide management of erythrocytosis due to congenital defects or in those with idiopathic erythrocytosis. Reducing the HCT by phlebotomy/venesectiondecreases the blood viscosisty and may be ofbenefit. Low dose aspirin may help to reduce thrombosis.

**Conclusions**

The above case is a rare case of congenital erythrocytosis. All causes of erythrocytosis should be kept in mind while evaluating a case of erytrocytosis. Since there are many causes of erythrocytosis an algorithmic and reflexive testing strategy is useful and serum EPO levels serve as an initial guide to further investigations.