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Concomitant Iron Deficiency Anaemia and Beta Thalassaemia Trait – A Case Report

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Abstract
Iron deficiency anaemia (IDA) often coexists with or superimpose on Beta thalassaemia trait(BTT), causing dilemma in diagnosis and management strategy. Iron deficiency frequently causes a low or normal HbA2 in HPLC chromatogram (5) thereby missing the diagnosis of concomitant BTT.
Correction of iron deficiency unmasks the concurrent BTT & subsequent HPLC reveals a supranormal HbA2, with improvement in anaemia.
Keywords: *IDA, BTT, HbA2 level before and after iron therapy.*

Case Report

30yrs female presented with chronic anaemia, mild icterus,and mild hepatosplenomegaly.

There was no lymphadenopathy, beeding disorder, joint pain or history of blood transfusion.

Investigations revealed the following.

Hb 5.8 gm/dl, RBC 3.07 millions/cmm, PCV 21.0%, MCV 68.3 fl, MCH 19 pg, MCHC 27.9 gm/dl, RDW CV 19.9% & Reticulocytes 3.9%. TLC, DLC and platelets were within normal limits.

Peripheral smear showed mild anisopoikilocytosis, hypochromia, microcytosis, few target cells, dacryocytes and occasional polychromatophils, (Fig.1, 2, 3).

Serum iron level was 40mg/dl, TIBC 485mg/dl, Transferrin saturation 8.2% & Serum Ferritin 6 ng/dl (low iron, low ferritin, low TSAT and high TIBC).

Hemolytic work up revealed a raised serum bilirubin, dominantly unconjugated fraction (total 2.7 mg/dl, unconjugated 2.0 mg/dl), diminished osmotic fragility (mean corpuscular fragility 0.30%), negative DCT and negative Sickling tests, low serum haptoglobin (less than 10mg/dl(ref. range : 30 - 200), non defficient G6PD, and a normal HPLC pattern of Hemoglobin with HbA2 : 3.0% (Fig. 4).

Considering possibility of concurrent sideropenia and a haemolytic condtion (hypochromic microcytic blood picture with low serum iron, low fertitin, low TSAT, as well as reticulocytosis, unconjugated hyperbilirubinaemia and low serum haptoglobin), patient was administered oral iron supplement for 16 weeks.

Subsequent follow up showed clinical improvement of anemia and a repeat HPLC revealed a supranormal HbA2 level of 4.6% (Fig 5). Red cell indices, haemoglobin, Serum iron & Ferritin increased, with reduction in TIBC.

With high index of suspiscion & relevant parameters, patient was diagnosed as BTT with coexistent IDA.

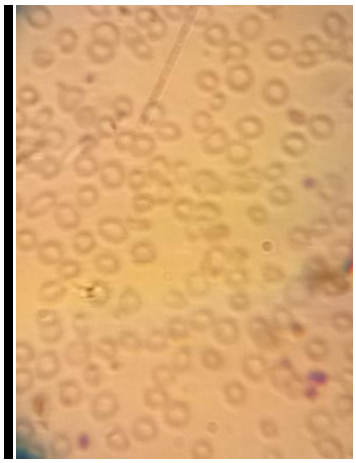

Fig - 1

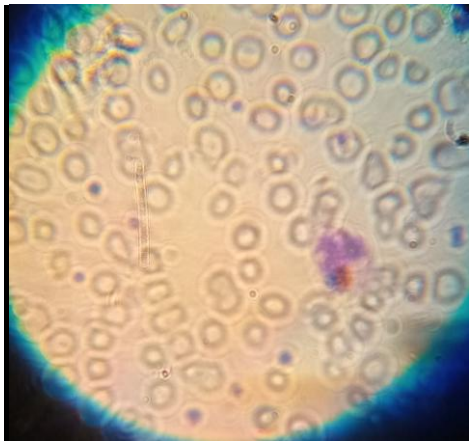

Fig – 2

Fig - 3

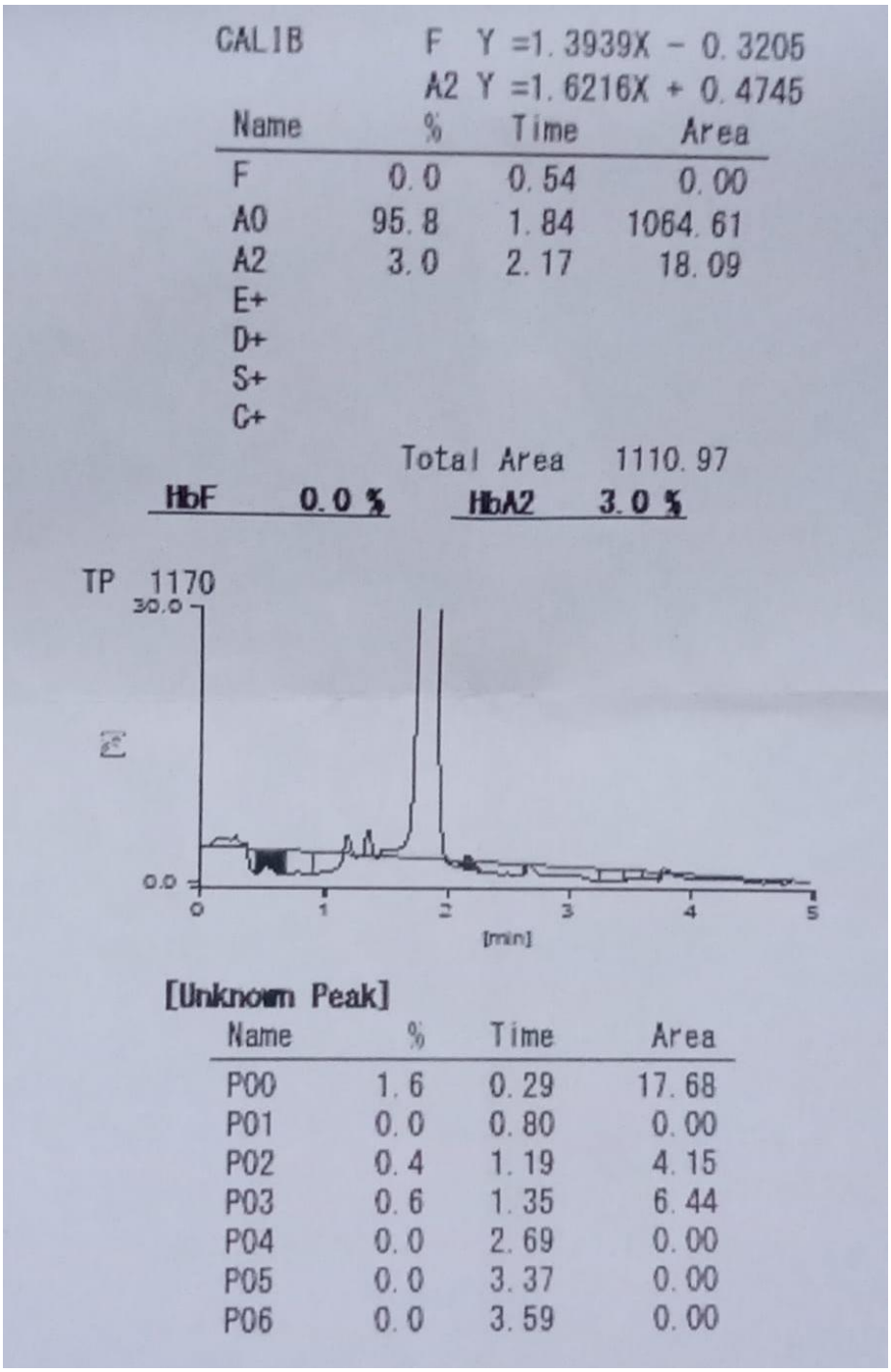

Fig – 4 HPLC (Before Iron Therapy

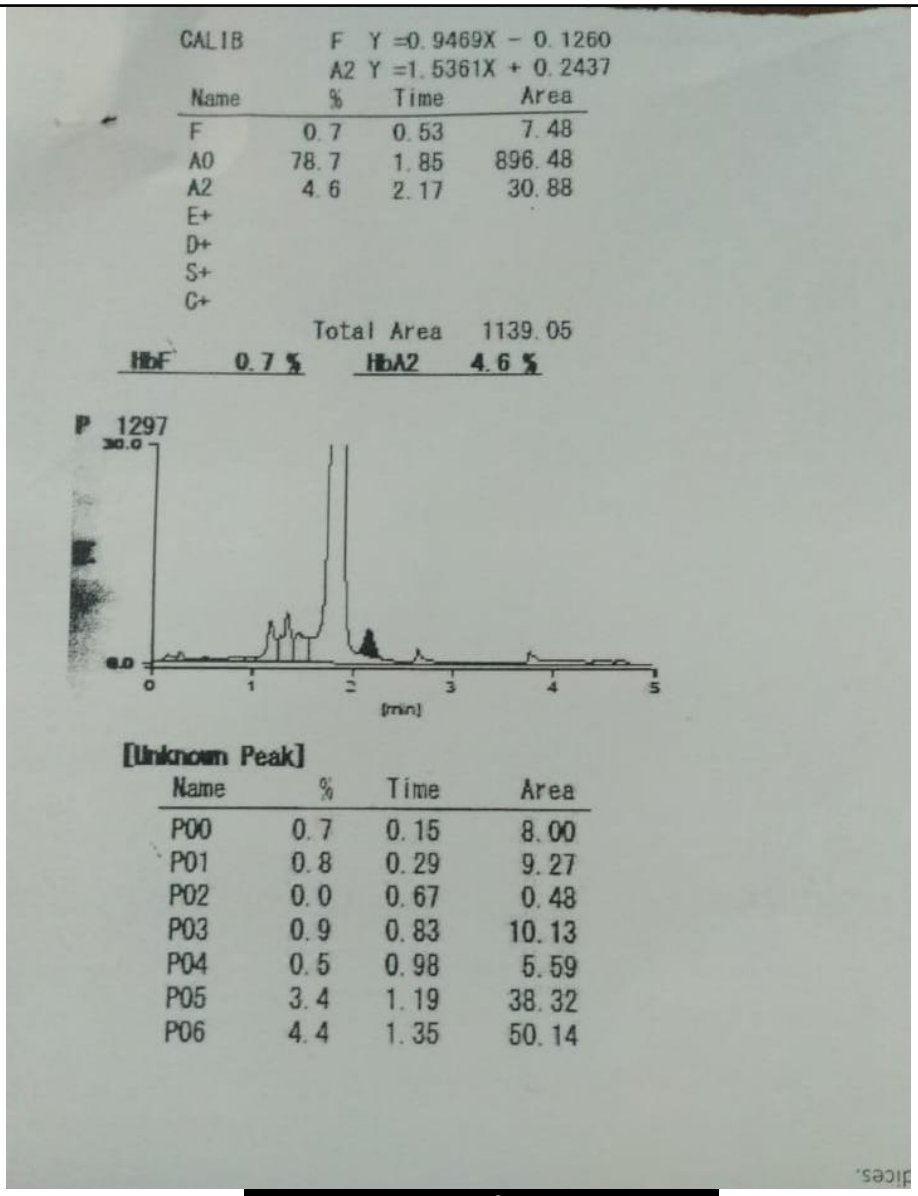

Fig – 5 HPLC (After Iron Therapy

Discussion

Both iron deficiency and beta thalassaemia trait are common in indian population. BTT occurs in 3.5%to 10% of Indians, while IDA is found in 70% - 80% of children, 80%pregnant women and 24% of adult male[1]. Both conditions show hypochromic microcytic anemia, and can coexist frequently.

Iron deficiency in presence of BTT, often leads to low or normal HbA2 level in HPLC, thus masking diagnosis of BTT, and creating diagnostic dliemma. If one such missed BTT individual gets married to another BTT, there is risk of birth of major thalassaemic children. In one study[2] HbA2 is low in 93.7%, normal in 2.4%, and raised in 4.4% cases of concurrent BTT with iron deficiency.

Suppression of HbA2 level by low iron is thought to be caused by defective synthesis or impaired binding of delta chain with heme, though some school suggests alpha chain abnormalities caused by low serum iron [4]

Proper diagnosis of iron deficiency and oral iron supplementation, for a recommended period of 16 weeks [3] are advised.

Post iron therapy investigations reveal clinical improvement of anaemia & increase in red cell indices, rise in haemoglobin, ferritin, iron and reduction in TIBC, along with significant increase in HbA_2 in HPLC.

Relation between hemoglobin level, RBC count and Mentzer index (PCV/RBC) is often used to differentiate probable IDA from probable BTT, in cases with microcytic hypochromic blood pictures. Reduced RBC level proportionate to haemoglobin and higher Mentzer index (above 13) suggest probable IDA, while disproportionate increase in RBC count in relation to haemoglobin and a low Mentzer index (below 13) favour a probability of BTT.

But this generalisation often fails when both IDA and BTT coexist.

Conclusion

Concomitant IDA and BTT is common, mostly in children, adolescents and pregnant women, often causing missed diagnosis of BTT, due to spuriously low or normal HbA2. This carries risk of having major thalassaemic children in case one such individual marries another BTT.

Clinical awareness leding to a proper diagnosis is of utmost importance in all anaemias in pregnancy & in anaemic children / adolescents.

This case is reported to highlight not so uncommon incidence of such "coexistence", which by no means can be called "peaceful".

Conflict of Interest: None.

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