

CASE REPORT

A case of concomitant iron and folate deficiency in a 26-year-old female with beta thalassemia minor.

Muhammad Irtza Tanveer¹, Saima Mansoor Bugvi², Sadaf Farzand³, Areeba Manzoor⁴, Ambereen Anwar⁵

ABSTRACT... We are reporting a known case of a 26-year-old lady with beta thalassemia minor (also called as beta thalassemia trait) who developed severe anemia due to co-existing iron and folate deficiency. The patient showed hypochromic microcytic anemia with markedly reduced mean corpuscular volume (MCV), mean corpuscular hemoglobin (MCH), along with biochemical features of iron depletion and folate deficiency. The baseline laboratory evaluations showed low values of Hb, serum iron, serum ferritin and transferrin saturation, along with increased TIBC. The patient was treated with intravenous ferrous sulfate (200 mg per week for 4 weeks), oral ferrous sulfate (14 mg/day for 10 weeks), oral folic acid (5 mg daily for 10 weeks), and intramuscular methycobalamin (500 mcg for 7 doses on alternate days). By the end of the 10th week patient showed a significant improvement in haematological and biochemical parameters. This case highlights the need for screening and treating the nutritional deficiencies in patients with beta thalassemia minor to prevent the worsening of anemia.

Key words: Beta Thalassemia, Folate Deficiency, Iron Deficiency.

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INTRODUCTION

Beta thalassemia minor is a heterozygous carrier state of beta-globin gene mutation, characterized by reduced beta-globin chain synthesis, microcytosis, and mild anemia. Most carriers remain asymptomatic; however, anemia worsens when concomitant nutritional deficiencies, particularly iron or folate deficiency, occur. Iron deficiency is common in females of reproductive age group due to menstrual blood loss.¹ Beta thalassemia minor females are also at risk of developing iron deficiency particularly in their reproductive years. There are studies showing that iron deficiency and beta thalassemia minor coexists and around 33% of the beta thalassemia minor patients are at risk of developing iron deficiency.² Folate deficiency may also develop in hemolytic states such as thalassemia due to increased erythropoietic activity.³ The coexistence of these deficiencies in thalassemia carrier patients complicates diagnosis and management because microcytosis is a feature common in both conditions, but their therapeutic approaches differ.

DISCUSSION

A 26-year-old married female with a known diagnosis of beta thalassemia minor and mother

of a beta thalassemia major girl presented in our Out-patient Department with history of generalized weakness, exertional fatigue, and numbness in her hands and feet for three months. She had no history of overt bleeding, chronic illness, or recent infections. Menstrual cycles were regular with moderate flow. On examination, she was pale but hemodynamically stable. There was no sign of icterus, lymphadenopathy, or organomegaly.

Baseline laboratory evaluation showed hemoglobin (Hb) 9.8 g/dL, hematocrit (HCT) 32.1%, mean corpuscular volume (MCV) 51.9 fL, mean corpuscular hemoglobin (MCH) 16.0 pg, and red cell distribution width (RDW) 17.4%. Total leukocyte count was $10.7 \times 10^9/L$, and platelet count was elevated at $487 \times 10^9/L$ indicating reactive thrombocytosis. Iron studies revealed severe deficiency: serum iron 22 $\mu\text{g}/\text{dL}$, total iron-binding capacity (TIBC) 559 $\mu\text{g}/\text{dL}$, transferrin saturation 3.39%, and serum ferritin 3.0 ng/mL. Serum folate was markedly reduced at 0.396 $\mu\text{g}/\text{dL}$, while vitamin B12 was in the low-normal range at 309 pg/mL. HbA₂ was elevated to 5.9%, confirming beta thalassemia

1. BSc. (Hons), MLT Trainee Pathology, Punjab Institute of Cardiology, Lahore.

2. MBBS, M.Phil (Haematology), FCPS (Haematology), Consultant Haematology, Punjab Institute of Cardiology/ Noor Thalassemia Foundation.

3. MBBS, M.Phil (Pathology), Consultant Pathology, Punjab Institute of Cardiology/ Noor Thalassemia Foundation.

4. BSc (Hons), MLT, M.Phil (Molecular Biology) Pathology Technologist, Punjab Institute of Cardiology, Lahore.

5. MBBS, M.Phil (Pathology), Professor and Head Pathology, Punjab Institute of Cardiology.

Correspondence Address:

Dr. Saima Mansoor Bugvi
Department of Haematology, Punjab Institute of Cardiology/ Noor Thalassemia Foundation.
dr.saimamsoor@gmail.com

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minor. Reticulocyte count was 1.2%, and lactate dehydrogenase (LDH) was also in normal range at 209 U/L. Her peripheral blood smear showed severe hypochromic, microcytic RBCs with marked anisopoikilocytosis and occasional macrocytes and the pictures of peripheral blood smear is shown in Figure-1. The patient was treated with intravenous ferrous sulfate 200 mg once weekly for four weeks, oral ferrous sulfate 14 mg daily and folic acid 5 mg daily for 10 weeks, and intramuscular vitamin B12 500 µg on alternate days for seven doses. Dietary advice for iron- and folate-rich foods was also given. At week 4, Hb increased to 10.2 g/dL, MCV to 55.6 fL, and RDW decreased to 16.6%. By week 10, Hb improved to 10.7 g/dL, MCV to 58.1 fL, RDW normalized to 14.1%, serum ferritin increased to 37 ng/mL, folate normalized to 8.4 µg/dL, and vitamin B12 rose to 423 pg/mL. Platelet count reduced to 349. All laboratory results of our patient at day 1, Week 4 and Week 10 are enlisted in table below.

In beta thalassemia minor, hypochromia and microcytosis is due to reduced beta-globin chain synthesis; however, concurrent nutritional

deficiencies can exacerbate anemia and cause more pronounced hematological abnormalities i.e., markedly low MCV and MCH. Our patient's severe iron deficiency was evident from very low iron and ferritin with elevated TIBC, and thrombocytosis while folate deficiency was confirmed by extremely reduced serum folate levels. Elevated RDW at presentation reflected anisocytosis due to mixed deficiency states, which improved as nutritional deficiencies were corrected.

A slight increase in MCV during treatment is expected because the underlying thalassemia minor continues to produce microcytic red cells despite iron and folate repletion. Folate deficiency is common in thalassemia carriers due to chronic hemolysis and increased red cell turnover, and folate supplementation is essential in order to support erythropoiesis and prevent megaloblastic changes.⁴ Vitamin B12 supplementation was also given to ensure that borderline levels did not limit hematological recovery and also because starting folic acid therapy alone can cause neural damage.⁵

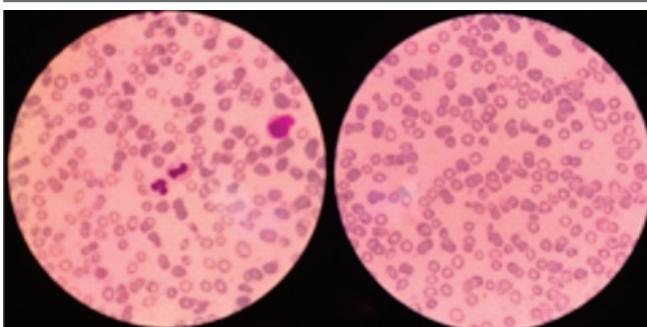
TABLE-I

Various hematological and biochemical parameters of subject at Day1, Week4 and Week10 respectively

Parameters	Day 1	Week 4	Week 10	Reference ranges
TLC	10.7	8.9	10.3	4-11 x10 ⁹ /L
RBC	6.18	6.0	5.91	3.8-5.8 x10 ¹² /L
Hb	9.8	10.2	10.7	11.5-16.5 g/dL
HCT	32.1	33.4	34.7	36-46 %
MCV	51.9	55.6	58.1	76-96 fL
MCH	16.0	17.1	18.2	26-32 %
RDW	17.4	16.6	14.1	12-16 %
Platelets count	487	421	349	150-400 x10 ⁹ /L
Retics Count	1.2			0.5-2.5%
HbA ₂	5.9			1.5-3.5%
Total Bilirubin	0.3			Less than 1.2mg/dL
LDH	209			Less than 228U/L
Serum Iron	22			50-170 µg/dL
TIBC	559			240-425 µg/dL
Transferrin saturation	3.39			20-50 %
Serum Ferritin	3.0	37	22-322 ng/mL	
Serum Folate	0.396	8.4	3-17 mcg/dL	
Serum B12	309	423	200-900pg/mL	

FIGURE-1

The peripheral blood smear of the patient showing hypochromia 2+, microcytosis 2+, marked anisopoikilocytosis and occasional macrocytes

**CONCLUSION**

Coexistent iron and folate deficiency in beta thalassemia minor patients can lead to severe anemia and complicate symptoms. This case report marks the need that comprehensive laboratory evaluation, including iron profile, folate, and vitamin B12, should be performed in thalassemia minor patients having moderate to severe anemia to better treat their symptoms. Timely correction with appropriate supplementation can restore hematological parameters and improve patient outcomes.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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AUTHORSHIP AND CONTRIBUTION DECLARATION

1	Muhammad Irtza Tanveer: Conceptualization of study, Data collection.
2	Saima Mansoor Bugvi: Study Design.
3	Sadaf Farzand: Literature search.
4	Areeba Manzoor: Data collection.
5	Ambereen Anwar: Proof reading.