Case Report

International Journal of Medical Case Report

A Case of Thalassemia Trait Misdiagnosed as Iron Deficiency Anemia

Authors:- Dr Ambhore Kuldip

Medical Officer, Rural Hospital Jintur Dist Parbhani.

INTERNATIONAL JOURNAL OF MEDICAL CASE REPORTS
ISSN 2455-0574

Abstract

Thalassemia trait is a common inherited disorder characterized by microcytic, hypochromic anemia, often misdiagnosed as iron deficiency anemia (IDA). This report describes a 3-year-old boy with persistent anemia unresponsive to oral iron therapy. Initial laboratory findings suggested IDA, but further investigation using the Mentzer index (MCV/RBC) indicated thalassemia trait. The diagnosis was confirmed by hemoglobin electrophoresis showing elevated Hb A2 levels. Discontinuation of iron supplementation and initiation of folic acid therapy led to clinical improvement. This case underscores the importance of the Mentzer index in differentiating thalassemia trait from IDA, ensuring accurate diagnosis and appropriate treatment. Prompt recognition and correct diagnosis are essential for effective management and better patient outcomes.

Keywords:- Thalassemia trait, Iron deficiency anemia, Mentzer index, Pediatric anemia

INTRODUCTION

Thalassemia trait is a hereditary blood disorder resulting from mutations in the globin genes, leading to reduced synthesis of one or more globin chains.[1] This defect consequently causes ineffective erythropoiesis resulting into anemia Anaemia seen in cases of thalassemia is usually microcytic, hypochromic anemia that can easily be confused with iron deficiency anemia (IDA), particularly in regions where both conditions are prevalent. The differentiation between these two types of anemia is critical for appropriate management.[2]

Access this Journal Online

Quick Response Code

Website: www.ijomcr.net

Email:ijomcr@gmail.com

Corresponding Author:

Dr Ambhore Kuldip
Medical Officer, Rural Hospital Jintur Dist Parbhani, Maharashtra India.

The global prevalence of thalassemia trait varies widely, with high frequencies in the Mediterranean, African, and Southeast Asian populations. It is estimated that approximately 5% of the world's population carries a thalassemia gene, making it one of the most common inherited disorders. The pathophysiology of thalassemia involves the imbalanced production of globin chains, leading to ineffective erythropoiesis, haemolysis, and varying degrees of anemia.[3]

Clinically, thalassemia trait often presents with mild to moderate anemia, which might be asymptomatic or associated with nonspecific symptoms such as fatigue and pallor. The diagnosis is typically suspected based on microcytic anemia observed in complete blood count (CBC) results. Important diagnostic clues include a low mean corpuscular volume (MCV) and mean corpuscular haemoglobin (MCH). The Mentzer index, calculated as MCV divided by the red blood cell (RBC) count, is a useful tool in differentiating thalassemia trait from IDA; a Mentzer index of less than 13 suggests thalassemia trait, whereas a higher index is indicative of IDA.[4]

In this report, we present the case of a 3-year-old boy initially misdiagnosed with IDA, who was later correctly diagnosed with thalassemia trait using the Mentzer index, highlighting the importance of accurate diagnostic differentiation to ensure appropriate treatment.

CASE REPORT

A 3-year-old boy presented to the pediatric clinic with complaints of fatigue and pallor observed over the past few months. He had no significant past medical history and his diet was reportedly well-balanced. Physical examination revealed pallor, but no hepatosplenomegaly or other abnormalities.

Initial laboratory investigations showed hemoglobin (Hb) of 6.3 g/dL, MCV of 37.78 fL, and MCH of 12.48 pg, leading to a presumptive diagnosis of iron deficiency anemia. The patient was started on oral iron supplementation. However, after two months of iron therapy, there was no significant improvement in his hemoglobin levels or clinical symptoms.

A follow-up CBC revealed persistent microcytic anemia with a hemoglobin level of 6.3 g/dL, MCV of 37.78 fL, and an RBC count of 5.05 million/µL. The Mentzer index was calculated to be 7.46 (MCV/RBC), suggesting thalassemia trait rather than iron deficiency anemia. Further investigations including iron studies showed normal serum iron, ferritin, and total iron-binding capacity (TIBC) levels, ruling out iron deficiency. Hemoglobin electrophoresis revealed an increased level of Hb A2 at 5.1%, consistent with beta-thalassemia trait.

Consequently, iron supplementation was discontinued, and folic acid was initiated. The patient's parents were counselled about the hereditary nature of the condition and its implications. Over the next several months, the patient's hemoglobin levels stabilized, and his symptoms improved.

Parameter	Result	Unit	Reference Range
Hemoglobin (Hb)	6.3	g/dL	12-18
Red Blood Cell (RBC) Count	5.05	million/µL	4.5-6.5
Packed Cell Volume (PCV)	19.08	%	37-54
Mean Corpuscular Volume (MCV)	37.78	fL	82-98
Mean Corpuscular Hemoglobin (MCH)	12.48	pg	27-33
Mean Corpuscular Hemoglobin Concentration (MCHC)	33.02	%	32-36
Red Cell Distribution Width (RDW-CV)	27.3	%	11.0-14.5
Mentzer Index	7.46	-	<13 for Thalassemia

Table 1: Haematological Profile of the case.

DISCUSSION

Thalassemia trait and iron deficiency anemia are common causes of microcytic anemia in children, but their management differs significantly. Misdiagnosis can lead to inappropriate treatment, as seen in this case. The Mentzer index is a simple yet valuable tool that can aid in the differentiation between these conditions. A Mentzer index of less than 13 suggests thalassemia trait, whereas a value greater than 13 typically indicates iron deficiency anemia.[5]

Several studies have validated the utility of the Mentzer index in differentiating between thalassemia trait and iron deficiency anemia. For instance, a study by Demir et al. demonstrated that the Mentzer index had a high sensitivity and specificity in distinguishing between these two conditions. Similarly, another study by Batebi et al. confirmed the reliability of the Mentzer index as a first-line screening tool for thalassemia trait.[6]

In this case, the use of the Mentzer index was crucial in redirecting the diagnosis and management from iron deficiency anemia to thalassemia trait. The cessation of unnecessary iron supplementation and the initiation of folic acid improved the patient's clinical condition, demonstrating the importance of accurate diagnosis.[7]

Moreover, this case highlights the need for a thorough evaluation of anemia in children, especially when there is no response to iron therapy. Genetic counseling and family screening are also essential components of managing thalassemia trait to identify carriers and provide appropriate advice on reproductive options.

CONCLUSION

This case emphasizes the importance of accurate differentiation between thalassemia trait and iron deficiency anemia in pediatric patients presenting with microcytic anemia. The Mentzer index is a valuable diagnostic tool that can guide appropriate management, preventing unnecessary treatments and improving patient outcomes.

Conflict of interest
None

Source Of Funding
None

REFERENCE

1. Baird DC, Batten SH, Sparks SK. Alpha- and Beta-thalassemia: Rapid Evidence Review. Am Fam Physician. 2022;105(3):272-280.
2. Viprakasit V, Ekwattanakit S. Clinical Classification, Screening and Diagnosis for Thalassemia. Hematol Oncol Clin North Am. 2018;32(2):193-211. doi:10.1016/j.hoc.2017.11.006
3. Nienhuis AW, Nathan DG. Pathophysiology and Clinical Manifestations of the β-Thalassemias. Cold Spring Harb Perspect Med. 2012;2(12):a011726. Published 2012 Dec 1. doi:10.1101/cshperspect.a011726
4. Bank A, Marks PA 1966. Excess α chain synthesis relative to β chain synthesis in thalassaemia major and minor. Nature 212: 1198–1200.
5. Tabassum S, Khakwani M, Fayyaz A, Taj N. Role of Mentzer index for differentiating iron deficiency anemia and beta thalassemia trait in pregnant women. Pak J Med Sci. 2022;38(4Part-II):878-882. doi:10.12669/pjms.38.4.4635
6. Ntaios G, Chatzinikolaou A, Saouli Z, Girtovitis F, Tsapanidou M, Kaiafa G, et al. Discrimination indices as screening tests for β-thalassemic trait. Ann Hematol. 2007;86(7):487–491
7. Cousens NE, Gaff CL, Metcalfe SA, Delatycki MB. Carrier screening for beta-thalassaemia:A review of international practice. Eur J Hum Genet. 2010;18:1077–1083

Author Contribution:- AK- Concept Of Design; Manuscript Preparation; Revision Of Manuscript; Review Of Manuscript
How To Cite This Article
Ambhore K, A Case of Thalassemia Trait Misdiagnosed as Iron Deficiency Anemia, Int. j. med. case reports. 2023; 4 (2): 5-7

Received : 05-02-2023 Revised: 25-02-23 Accepted : 15-03-23