Original article:

Evaluation of blood indices and peripheral smear examination in beta thalassemia patients

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ABSTRACT:

Introduction: There are certain conditions with genetic significance which may affect the life of people even with no any symptoms. One of the most common examples is thalassemia. It is a quantitative hemoglobinopathy with the reduced synthesis of the globin chains of haemoglobin. It is one of the major health related challenges faced by the society.

Material and methods: Study was done at Central clinical laboratory, Department of Pathology, PDU Medical College Rajkot. Study participants were those who were admitted in PDU hospital as well as known cases of thalassemia major. Blood samples collected in EDTA(Ethylene diamine tetra acetic acid) vaccutte were tested in the automated cell counter and peripheral smear examination was done from the slides stained in Romanowsky stains.

Results: Automated cell counters and peripheral smear examination revealed many abnormalities in the blood indices and red blood cells lineage which helped to differentiate among thalassemia major, thalassemia minor as well as iron deficiency anaemia.

Conclusion: This study shows the importance of peripheral smear examination as a primary step in suspicion of diagnosis of thalassemia trait patients and iron deficiency anemia patients. Also the haemoglobin and red blood cell abnormalities in thalassemia major patients are described. These help the clinicians to monitor their blood transfusion requirements in these patients as well as to screen the heterozygous patients so as to reduce the birth rates of thalassemia major patients.

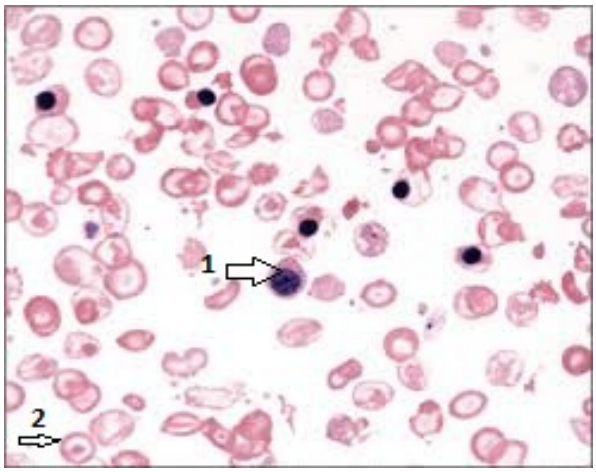
Keywords: Thalassemia, peripheral blood findings, red cell abnormalities.

INTRODUCTION: Anemia is one of the most common health related problems faced by the society. Although nutritional deficiency is the most common cause, other causes like hemolysis are also common. Hemolytic anemias are characterised by increased red cell destruction. Amongst the inherited causes of haemolytic anemias, thalassemias are the most common. Thalassemia is a quantitative hemoglo-binopathy due to reduced synthesis of one or more globin chains. β Thalassemia trait is generally an asymptomatic inherited condition with genetic significance only. In contrast thalassemia major is a significant transfusion dependant hemoglobinopathy that presents a major health challenge to even more developed and affluent countries. For majority of patients, lifelong transfusion remains the only treatment option which itself is associated with comorbidities and complications. The pressure on the healthcare resources including maintaining a safe blood supply is especially challenging. Early detection and characterization of thalassemia trait in community enables earlier intervention and

management in the hope of reducing the births of babies with thalassemia major, thereby reducing the psychological and emotional impact on the family and the society. Both thalassemia major and trait reveal different red blood cell abnormalities on peripheral smear examination. This could help to suspect these hemoglobinopathies and differentiate them from iron deficiency anemias.

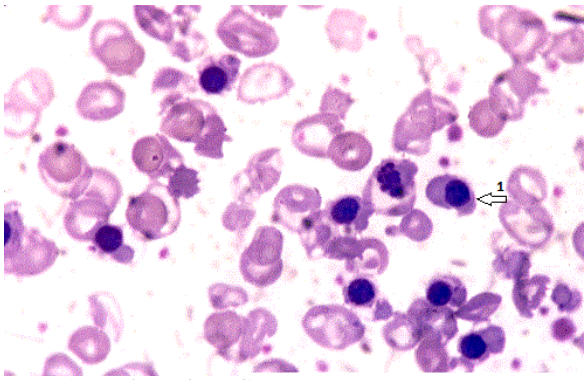
MATERIAL AND METHODS: Study was conducted in Central Clinical Laboratory, Department of Pathology, PDU Medical College Rajkot. Study participants were those admitted in PDU hospital, Rajkot as well as the known cases of β-thalassemia major. Participants of the study were the subjects with the age group 3 -30 years. Blood samples sent in EDTA(Ethylene diamine tetra-acetic acid) vaccutte were tested in automated cell counter SYSMEX KX 21 as well as peripheral smear examination was done from the slides stained in Romanowsky stains.

OBSERVATION AND RESULTS: 200 samples of the blood were selected for study. Out of them 100 samples selected were of known cases of β thalassemia major. Rest were of thalassemia trait and iron deficiency anemia. Following were the results.

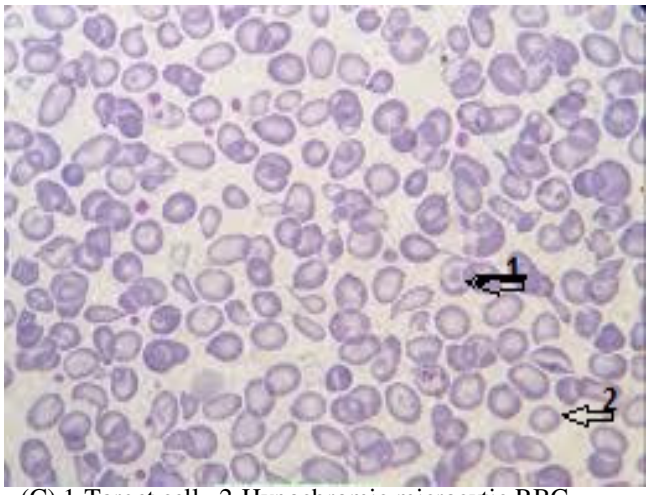


(A)

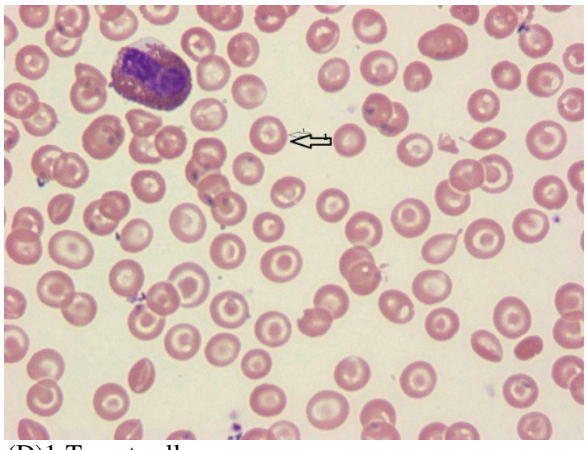
1 – Nucleated RBC, 2 – Target cell



(B) 1-Nucleated RBC



(C) 1-Target cell, 2-Hypochromic microcytic RBC



(D)1-Target cell

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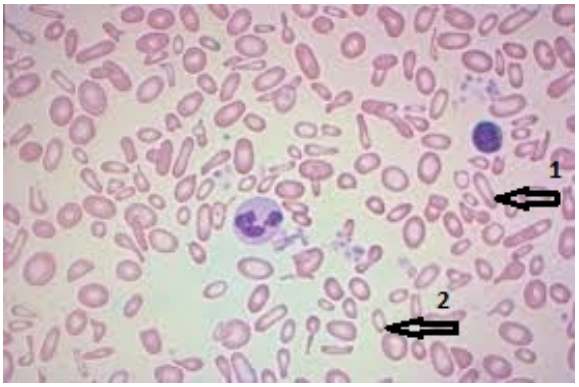
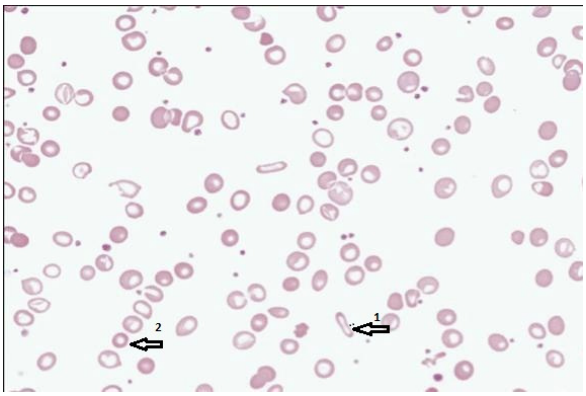


(E)1-pencil cell, 2-hypochromic microcytic RBC



(F) 1-Pencil shaped cell,2-Hypochromic microcytic RBC

Figures A and B show the commonly observed peripheral blood findings in thalassemia major in our study:

Anemia is moderate to severe, MCV (Mean Corpuscular Volume), MCH(Mean Corpuscular Hemoglobin),MCHC (Mean Corpuscular Hemoglobin Concentration) are reduced. Moderate to marked degree of anisopoikilocytosis is evident with hypochromic microcytic red cells. Target cells and many nucleated red cells are seen.

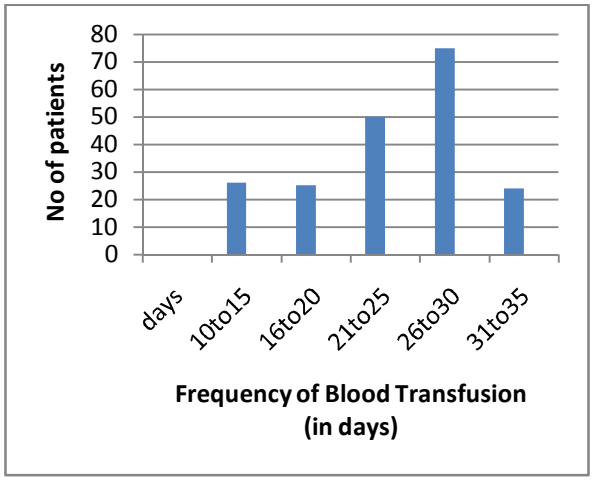
Figures C and D show the commonly observed peripheral blood findings in β thalassemia traits in our study.

Hemoglobin may be normal or slightly reduced. Red cell count is elevated. MCV and MCH are reduced. MCHC may be normal. RBC mass seems to be increased with mild degree of anisopoikilocytosis . hypochromic microcytic rbcs are seen with few target cells. Red cell distribution width (RDW) is normal to low. Figures E and F show the common peripheral blood findings in iron deficiency anemia in our study. Severe anemia with MCV, MCH and MCHC reduced. Peripheral smear shows mild to moderate degree anisopoikilocytosis. Red cells are microcytic hypochromic with few pencil shaped cells.

Following table shows the significant parameters of β thalassemia trait and iron deficiency anemia observed in our study by automated cell counter.[1]

Parameters	Thalassemia trait	Iron deficiency anemia
Hb	Decreased	Decreased
RBC COUNT	elevated	Decreased
MCV	Decreased	Decreased
MCH	Decreased	Decreased
MCHC	Normal	Decreased
RDW	Normal	Increased

Following graph shows the frequency of blood transfusion in thalassemia major patients in our study



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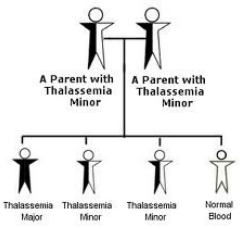
DISCUSSION:

Thalassemia is a genetically transmitted condition. It is autosomal recessive caused by decreased synthesis of the globin chains of haemoglobin.[2] To diagnose thalassemia at an early stage is of paramount importance for the health facilities to take steps for its prevention in the future generation. Our study was conducted amongst the known cases of thalassemia major and some incidental cases of thalassemia trait were found on the basis of peripheral smear findings and blood indices. Those were the cases whose samples were tested for routine blood count and peripheral smear examination done revealed the findings suspected of the β thalassemia trait which was confirmed later with the haemoglobin electr-ophoresis. Red cell indices, haemoglobin and peripheral smear examination provide the basic steps in screening of thalssemias and iron deficiency anemia. β thalssemia trait and iron deficiency anemias both have decreased MCV and MCH.[3] RDW is raised in iron deficiency anemia more than thalassemia trait. Also MCHC is low here. While β thalassemia trait has high RBC count and target cells in peripheral smear. This can help in the differential diagnosis of them. Thalassemia major is a transfusion dependant hemoglobinopathy. It poses a major psychological and emotional impact on the family and the society. The pressure on the health care resources including maintaining a safe blood supply is especially a challenge in country like India where its prevalence is high.



Thus early detection and characterisation of β thalassemia trait in community enables earlier intervention and management in the hope of reducing the number of births of babies with thalassemia major, reducing the mortality rates.[4] Screening for β thalassemia trait should be carried out at different stages. eg at school, premarriage, preconceptual or antenatal. Generally a presumptive diagnosis is made from the red cell indices, peripheral smear examinations, osmotic fragility tests, haemoglobin electrophoresis, etc. In most cases of thalassemia trait suspected from the peripheral blood examination in our study, haemoglobin A2 was between 4% to 8%. HbF was normal to slightly increased. Serum iron and ferritin were done in these patients which were found to be normal to increased because of increased absorption of alimentary iron. Patients with thalassemia major had increased iron and serum ferritin levels. Hb electrophoresis revealed HbF more than 90%. This were the confirmatory findings to our study. It is important to stress that the screening programmes provide a risk reduction but not a guarantee of protection or detection. Sometimes cases may be missed if the screening is done only on the basis of red cell indices that are unable to capture the target population if there is co existing liver disease or Vit B12 deficiency or patients with HIV therapy, which may show raised MCV and MCH

CONCLUSION: Thus as an initial step of early detection of thalassemia, red cell indices and peripheral smear findings provide very useful guide for the clinicians to request for other investigations to confirm the diagnosis of thalassemia. Also they direct the clinicians to monitor the blood transfusion requirements in thalassemia patients along with the symptoms. This helps to avoid frequent unnecessary blood transfusions in thalassemia major and thus to

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ensure them a safer and longer life. Methods used in screening programmes provide only a presumptive diagnosis and must be followed by DNA analysis techniques to arrive at a definitive diagnosis.[5]

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