Journal Of Harmonized Research (JOHR)

Journal Of Harmonized Research in Medical & Health Sci.
2(3), 2015, 68-76

ISSN 2395 – 6046

Original Research Article

PREVALENCE OF BETA-THALASSEMIA TRAIT IN AND AROUND PATNA, BIHAR

Dr. Iffat Jamal,

MD Pathology
Department of Pathology, Patna Medical College and Hospital, Patna, Bihar, India

Abstract: Beta-thalassemia continues to be a major burden to the society particularly in developing countries like India.Among its various types,prevalence of β-Thalassemia trait in India varies from <1% to 17% with an average of 3.3%.There is a diagnostic challenge in distinguishing it from Iron deficiency anaemia as both present with features of microcytic hypochromic anaemia. **Aim:** This study was carried out to assess the prevalence of β-Thalassemia trait in and around Patna so that a rough estimate could be made regarding its occurrence in patients presenting with chronic resistant microcytic anaemia. **Materials and methods:** The study was carried out in 300 patients coming to PMCH with complains of fatigue, weakness, lethargy and dizziness.Out of 300 cases, 197 cases were proved to be of Iron deficiency anaemia by their low serum ferritin level and peripheral blood picture. Rest 103 cases with normal or increased serum ferritin level were screened by NESTROF TEST, with 31 giving positive results which were then subjected to Haemoglobin electrophoresis .HbA2 was found to be raised(>3.5%) in 5 patients confirming the diagnosis of Beta-thalassemia trait in them. **Results:** Prevalence of β -thalassemia trait in the study sample was-5/300 =0.016 i.e. 1.6%. **Conclusion:** This study gave a rough estimate of prevalence of β-thalassemia trait in our region.Recognition of β-Thalassemia Trait is important in differentiating it from microcytic hypochromic anaemia of Iron deficiency origin.

Key words: β-thalassemia trait,electrophoresis,microcytic,hypochromic.

Introduction: The Thalassemic syndromes are a heterogenous group of inherited disorders caused by genetic defects leading to decreased synthesis of either α or β globin chain of Hb[1].Depending on which Hb chain which is deficient it is termed α-Thalassemia o β-Thalassemia β-Thalassemia is the commonest inherited haemoglobinopathy[2]. Depending upon the extent of decrease in β-globin chain synthesis and degree of clinical severity,it is further classified.βTT cases are heterozygous f or thalassemia gene &remain mostly asymtomatic.

For Correspondence:
iffatjamal111@gmail.com
Received on: August 2015
Accepted after revision: September 2015

It is estimated that 3 % of world's population are carriers of beta thalassemia . The South east asian region that also includes India accounts for 50% of the world carriers.In India nearly 30 million people are carriers of Beta - thalassemia . The prevalence of beta-thalassemia trait (βTT) in India has variously been reported from <1% to 17% with anaverage of 3.3%[3].Certain communities in India like Gujratis, Sindhis, Punjabis, Bengalis, Lohanas, Bhanusalis etc. have higher frequency of this trait.

Materials and Methods:

This study is a hospital based observational study conducted in the Deptt of Pathology, PMCH. It was carried out in 300 patients of different age groups coming to OPDs of various specialities with symptoms of weakness, lethargy, fatigue, dizziness, loss of appetite. Children & adults of both sexes of ages ranging from 3yrs to 60 yrs were included. A detailed clinical history was taken followed by Haematological investigations like Complete blood count, S.Ferritin & PBF to confirm anaemia & to sub-type it. Out of 300 pts, 197 cases were proved to be suffering from Iron deficiency anaemia by their low serum ferritin level & microcytic hypochromic peripheral blood picture. Rest 103 cases with normal to increased serum ferritin with microcytic hypochromic/normocytic normochromic peripheral blood pictures were screened by NEST ROF TEST, with 31 giving positive results which were then subjected to Haemoglobin Electrophoresis for confirmation of βTT .HbA2 was found to be raised (>3.5%) in 5 of them confirming the diagnosis of βTT in them.

Observations: Majority of pts (270/300) had following haemogram findings. Hb %, RBC count, MCV, MCH, MCHC decreased.RBC count and reticulocyte count was decreased to slightly elevate. RDW-increased to normal. Peripheral blood picture showed anisopoikilocytois with microcytosis, hypochromia, targetcells, NRBCs and basophilic stippling in few cases. 29 cases had all these parameters within normal range,served as control in this study. Then serum Ferritin levels were assayed in patients with microcytic hypochromic or normocytic normochromic peripheral blood pictures. 197 cases had low S.ferritin & rest had either normal or raised S.ferritin level.In the later group (normal to raised S.ferritin) NESTROFT was performed.31 cases turned out to be Nestrof test positive. These Nestrof test positive cases were then subjected to Hb Gel electrophoresis, in which out of 31 cases,5 cases had raised HbA2 level of >3.5% confirming the diagnosis of β-thalassemia trait in them.

Results:

- Maximum no. of pts belonged to 21-30 years of age group(94/300-31.33%).
- Majority of the patients were females of reproductive age group.M/F ratio-0.68:1
- Majority of the pts with NESTROFT +vity were in age group of 21-30 yrs.(17 /31)
- Overall prevalence of βTT in the study= 5/300=0.16 or 1.6%

AGE IN YEARS	MALE	FEMALE	TOTAL	+VE NESTROFT	RAISED HbA2(>3.5 %)
3–10	7	3	10	0	0
11–20	21	42	63	6	0
21–30	37	57	94	17	4
31–40	26	32	58	7	1
41–50	21	27	48	1	0
51–60	10	17	27	0	0
TOTAL	122	178	300	31	5

Table 1 showing age and sex wise break up of NESTORT +VE cases and cases with raised HbA2 level >3.5%.

Discussion:

- ❖ As per this study the prevalence rate of β-thalassemia Trait in & around Patna was 1.6%.
- This was in accordance with the previous study done by Madan et al.2010[4]
- The prevalence rate of βTT in our region was lower than that reported by Flatz *et al. in* Assamese population (5%),Mishra *et al.reported 8% prevalence of beta thalassaemia* trait among the coastal population of Orissa.
- It is higher in WB(3.97-4.9%),Gujrat(4.02-3.5%),Maharashtra(4.3-2.91%), Punjab(5.62-4.38%) as per the multicentre study done by Mohanty et al. [5]
- ❖ Thus it proves that βTT follows genetic,ethnic & geographical variations.
- ❖ Age and sex wise prevalence of βTT was similar to that observed by Mamtani et al.(2007) [6].
- ❖ NESTROT is an useful ,quick,cheap,less time consuming screening test for beta-thalassemia trait. Its abbreviated form of NAKED EYE SINGLE TUBE RED CELL OSMOTIC FRAGILITY TEST
  METHOD-5ml of 0.35% saline solution taken in 2 test tubes.In one tube 0.02ml of blood of normal person(control tube)& in other 0.02ml of pt's blood is taken(test tube). After ½ hr a white paper with a dark black line is placed behind both tubes In CONTROL TUBE-BLACK LINE clearly seen.
  In TEST TUBE-black line not clearly seen in +ve cases
  REASON-Microcytic hypochromic red cells are resistant to hemolysis in hypotonic saline solution & remain opaque[7].
- Gel Haemoglobin Electrophpresis: Gel Hb electrophoresis at an alkaline pH of 8.4 was performed for detection of βTT.
- PRINCIPLE:Due to –ve charge of Hb,Hb migrates from cathode to anode in alkaline medium.Changes in degree of –ve charges in diff types of Hbs allow separation of abnormal variants from HbA.
- In βTT Hb electrophoresis shows : HbA=90-93%,HbA2=3.6-8% DIAGNOSTIC,HbF=normal or slightly raised (1-4%)

<u>Recognition of Beta-Thalassemia Trait is important: WHY???</u>

1. In differentiating from microcytic hypochromic anaemia of Iron deficiency,so that unnecessary Iron therapy can be avoided which can lead to iron overload.

2.In Genetic counseling: of βTT carriers-Beta-thalassemia follows an autosomal recessive mode of inheritance[8],so if both parents have this trait, there is a 25 percent (1 in 4) chance **with each pregnancy of having a child with Beta Thalassemia Major ,**so by prior genetic counselling & PRENATAL DIAGNOSIS, parents can choose whether to continue the pregnancy or not.

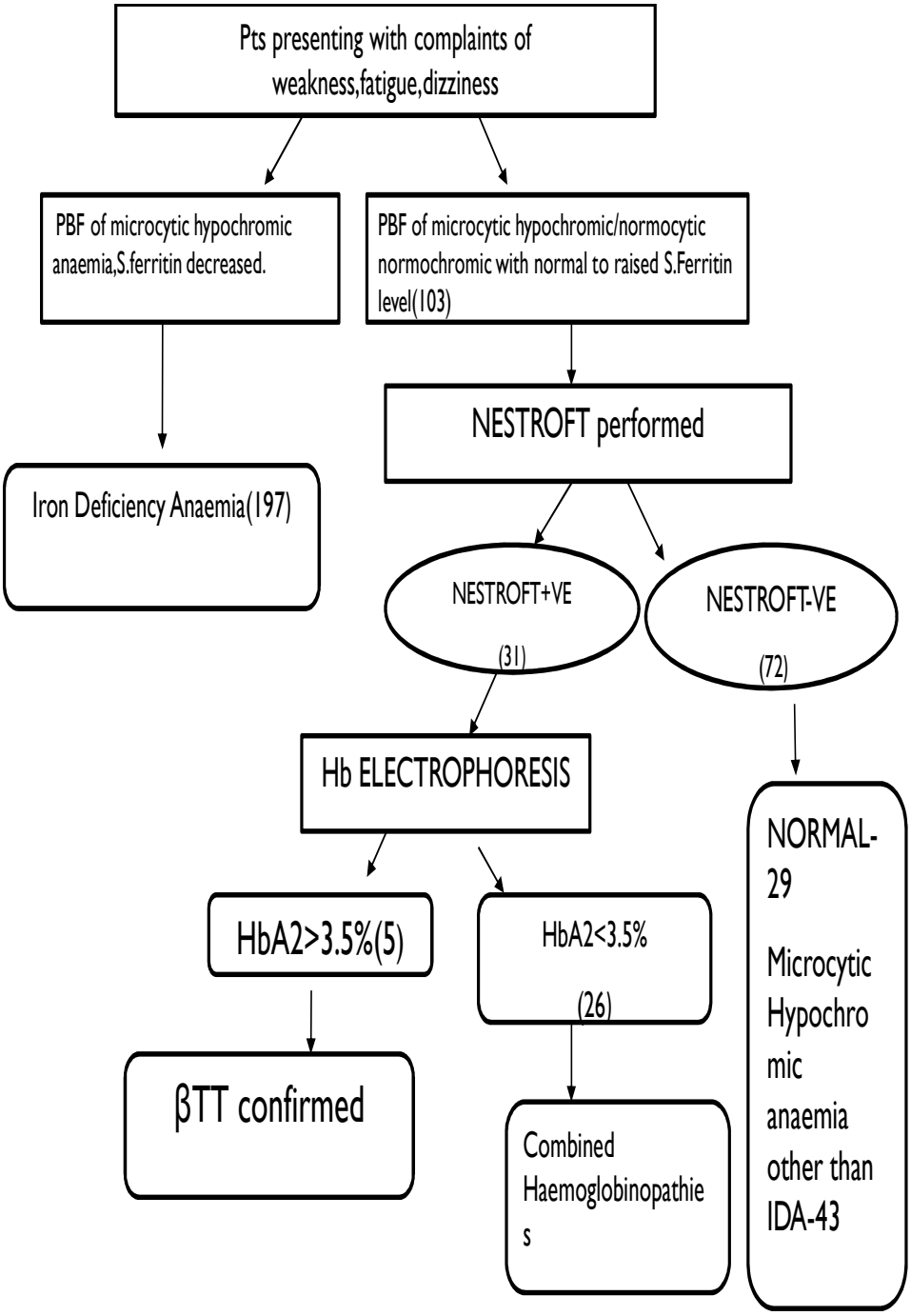

Figure-1- Flow chart showing the step-wise approach followed for this study.

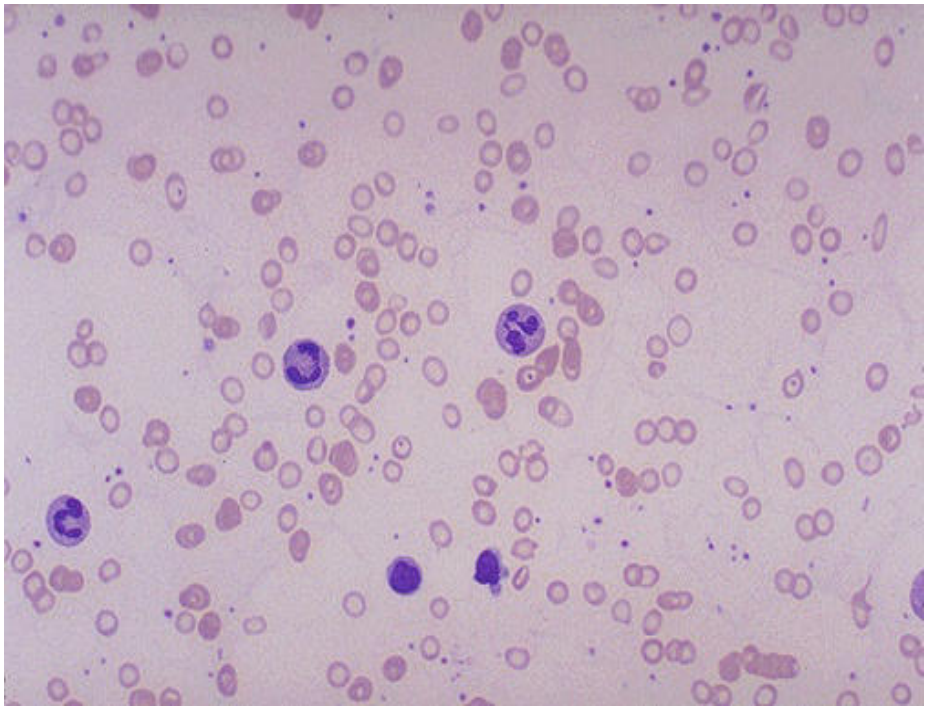

Figure-2- Peripheral blood picture showing microcytic Hypochromic red cells

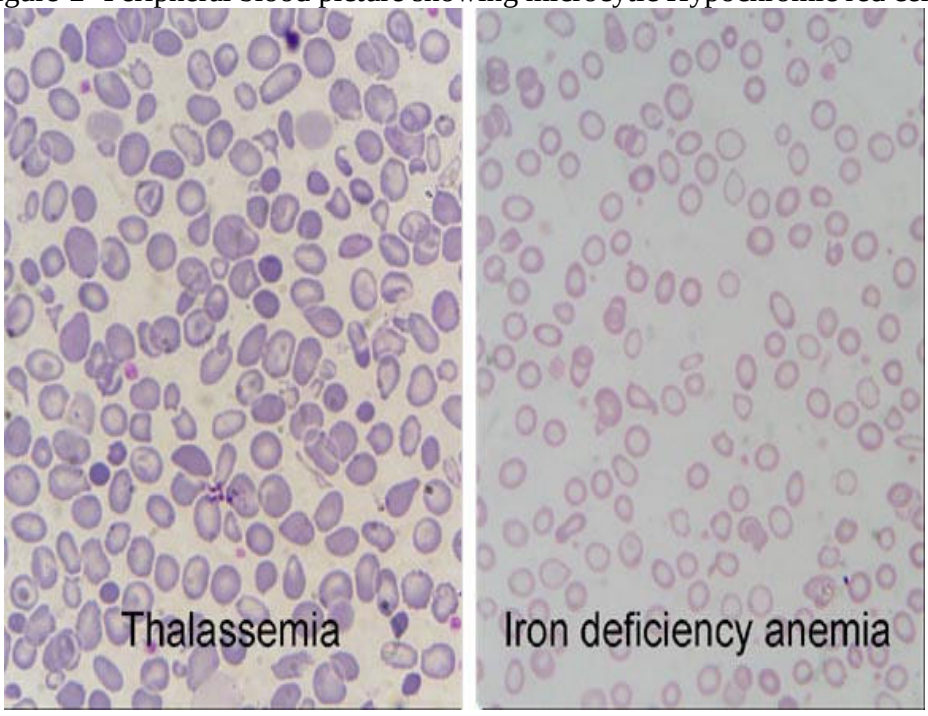

Figure 3- showing comparsion of peripheral blood picture in Thalassemia and Iron Deficiency Anaemia.

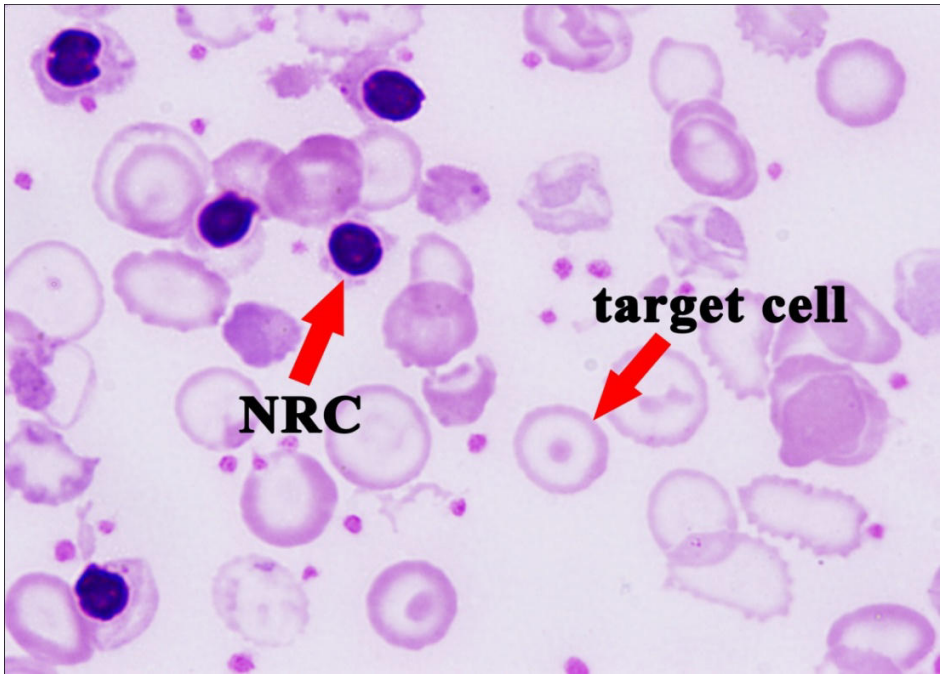

Figure-4- showing nucleated RBCs,target cells which are indicators of hemolytic anaemia.

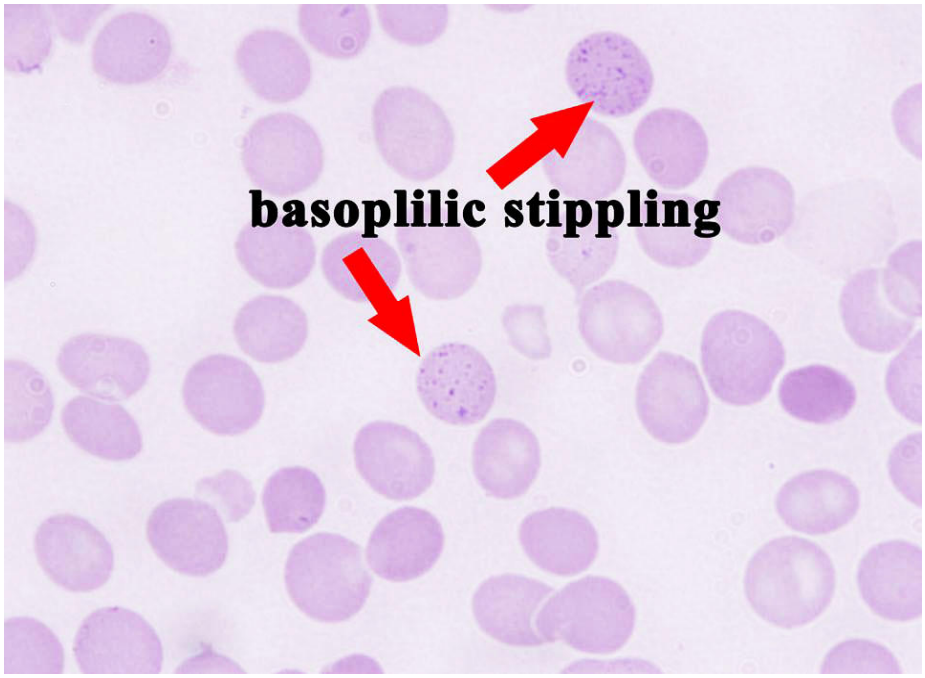

Figure 5- Peripheral Blood smear showing basophilic Stippling.

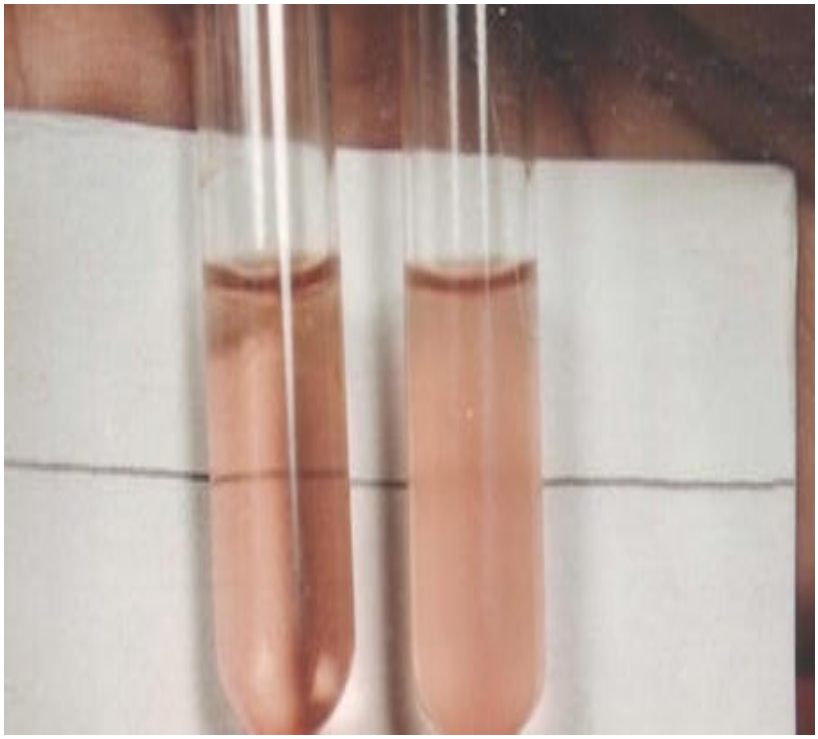

Figure 6- Figure showing Positive Nestrof test

Figure 7- showing Negative Nestrof Test.

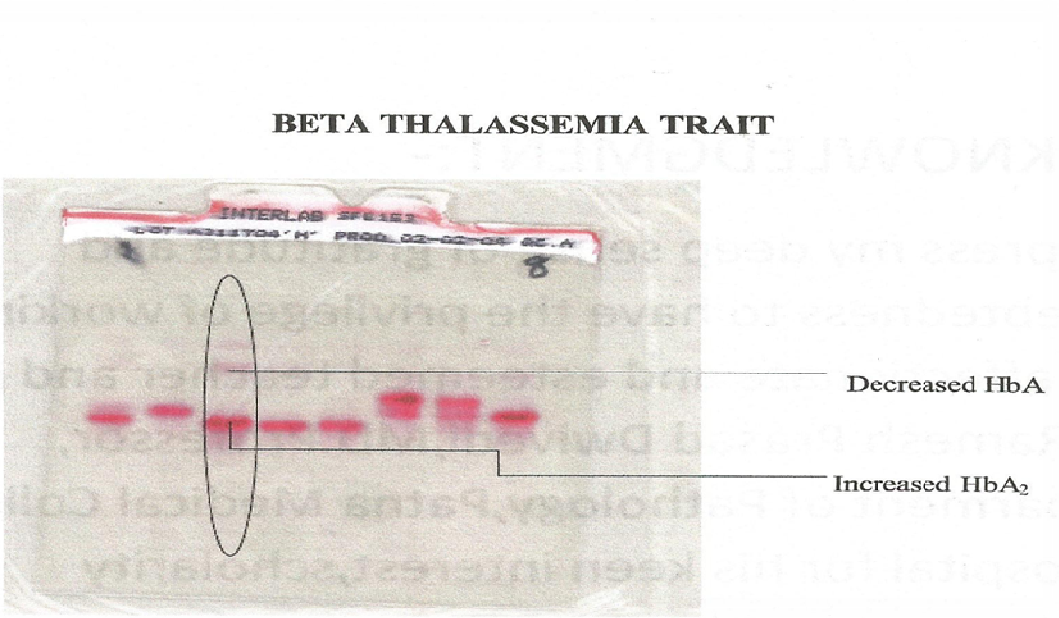

Figure 8- Hb Gel electropohoresis- showing broad band of Hb A2

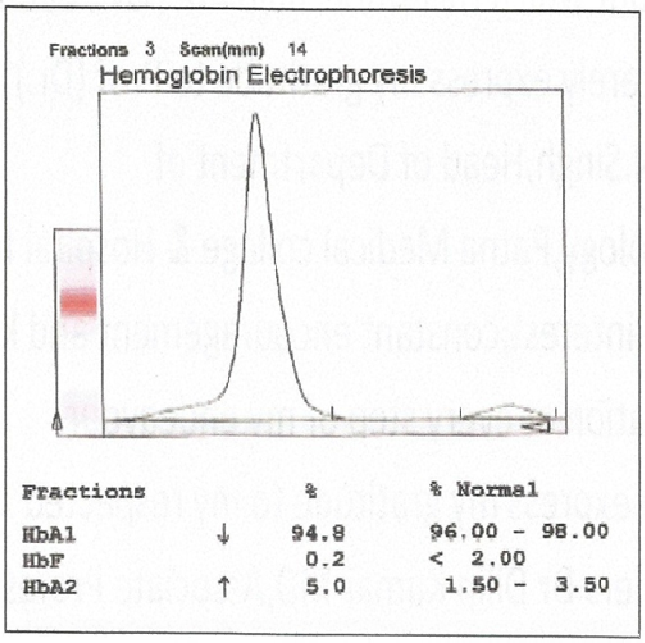

Figure 9- Graph showing raised HbA2 level confirming the diagnosis of Beta –thalassemia trait.

Conclusion:

1} Based on this study,prevalence rate of βTT in our region was 1.6%.

2} The haematological findings of IDA & βTT are quite similar,so this disease entity should always be kept in mind while dealing with cases of resistant microcytic hypochromic anaemia especially in βTT prevalent areas

3} Simple haematological tests like CBC, RBC indices,PBF analysis and Iron status studies can help in differentiating it from other causes of microcytic hypochromic anaemia & confirmed by Hb Electrophoresis,HPLC etc…

4} NESTROFT fits perfectly in the category of screening test for βTT-as it is simple,easy to perform,economical,doesn't require any sophisticated equipment & has high sensitivity & high –ve Predictive value[10].

5} Thalassemia is a major public health problem in certain communities in India,so a proper screening & Genetic counselling of βTT cases can prevent births of β-thalassemia major children ,for which an effective THALASSEMIA CONTROL PROGRAMME is all that is required today.

References:

1.John P.Geer,John Foerster,George M.Rodgers,Frixos Paraskevas,2009.12th edition,Wintrobe's Clinical Haematology;1085-1118

2. Dacie JV,Lewis SM,11th edition,Churchill Livingstone:2012,Practical Haematology

3.Tejinder Singh,3rd edition:2014;133-146

4. Nishi Madan, Satendra Sharma, S. K. Sood, Roshan Colah1, (Late) H. M. Bhatia ;Frequency of β-thalassemia trait and other hemoglobinopathies in northern and western India: Indian Journal of Human Genetics January-April 2010 Volume 16 Issue 1:17-23

5. D.Mohanty,S.k.Sharma,U Chaudhari.Prevalence of β-thalassemia and other haemoglobinopathies in six cities in India:A Multicentre Study.

6.Mamtani M,Das K,Jawahirani A.Is NESTROFT sufficient fo mass screening for β-thalassemia trait?J.Med.Screen;2007;14:169-73

7.Richard A.McPherson,Matthew R.Pincus,2012,22th edition,Henry's Clinical Diagnosis and Management by Laboratory Methods;576-89

8. Sreekanthaet al. Biochemical and pathological aspects of thalassemia International Journal of Research in Health Sciences. Oct–Dec 2013 Volume- 1, Issue-3,140-150

9.Madan N , Meera S , Satendra S, Usha Rusia,Kusum Kela.Red cells Indices and Discriminantfunctions in the detection of Beta – ThalassemiaTrait in a population with High prevalance of IronDeficiency Anemia. Indian J. Pathol. Microbiol1999; 42(1): 55-61.

10. Sanjay Piplani et al., NESTROFT - A Valuable, Cost Effective Screening Test for Beta Thalassemia Trait in North Indian Punjabi Population