Genetic Diagnosis and Detective Set Up In Patient and Carrier of Beta-Thalassemia by Reverse Slot - Blot Strip Assay

Naji T, Sirati Moghaddam P*, Azari A, Kheirvari Khezerloo J, Abniki M

***Abstract*—** Intoduction:Thalassemia is a disorder in the globin chain as a result , the hemoglobolin. Leads to reduced a ability to carry oxygen , anemia , splenomegaly,Bone deformity and the detection techniques for couples before marriage is.
Method:In the present study ,40 of 130 patients with Thalassemia that were Medical cooperate in this reaserch were selected as, was used reverse slot blot technique to indentify mutations in patients.Spss software was used to analye the results.
Results:The level of Hb electrophoresis strip method were respectively (MCV<80fl) , (MCH<27pg) and(HCT<40), the mean and standard deviation of Hb(mg/ml) were 9.2 and 1.24, MCV(fl) were 87.24 and 0.10 and MCH (pg/cell) were 20.98 and 3.63 and Hb A2 were 2.93 and 0.50 and HCT were 32.30 and 5.43 for both gender.
Conclusion:Revers slot blot technique or stript can be determine accurately and in less time , with out the need for sophisticated laboratory equipment the beta-Thalassemia mutations and patients. Amore reliable method is a reverse hybridization that can reduce false negative results.Increase the accuracy of the test, the test time reduction are merits of reverse hybridization**.**

Index Terms--- BetaThalassemia, Electrophoresis, Allele, Mutation , Hemogolobin.

I. INTRODUCTION

Thalassemia is the most common form of all inherited disorders of the red cell [1]. It is estimated that 70 000 children are born with various forms of thalassemia each year, and more than half of these births are affected by severe forms of β-thalassemia, of which the most common subgroup is hemoglobin (Hb) E β-thalassemia [2,3]. Thalassemia was originally confined to the tropical and subtropical regions of the world, distributed throughout the Mediterranean, sub-Saharan Africa, the Middle East, and the southern regions of Asia. However, as a result of relatively recent migration, many north European and North American countries are now home to large numbers of patients with thalassemia [4]. The β-thalassemias are a group of hereditary hematological diseases caused by over 300 mutations of the adult β-globin gene [5] with excellent reviews providing background information outlining genetics [6,1] pathophysiology [7,8], and therapeutics [9] of β-thalassemia that is beyond the scope of this review. In brief, β-thalassemia is brought about by mutations reducing or abrogating β-globin expression, which thus lead to reduced adult hemoglobin ([HbA] an $\alpha_2\beta_2$ heterotetramer) and excess α-globin content in erythroid cells, in turn resulting in ineffective erythropoiesis and apoptosis in the erythroid lineage [10]-[12]. Most β-thalassemia patients therefore require lifelong clinical management by blood transfusion and chelation therapy [13,14] with a few having the option of curative but potentially hazardous allogeneic transplantation of hematopoietic stem and progenitor cells (HSPCs) instead [15,16]. This indicates the need for alternative therapies, and the observation that high levels of the fetal β-globin-like γ-globin chain result in an ameliorated β-thalassemia phenotype [17] has prompted the search for γ-globin-inducing chemical agents [18-19] .Patient response to known γ-globin inducers, however, is varied [20] and the search continues for reagents with higher efficiency, consistency, and tolerability in chronic application [21] if not to cure the disease, then to reduce transfusion requirements and the significant cost of disease management.

II. MATERIAL AND METHODS

This type of research was applied and basis. Statistical Society was composed of Patients with major thalassemia. Samples Patients referred to Children's Medical Center Blood samples were studied in 2012-2013 years. In Sampling, fertility center of Imam Khomeini hospital in medical records about 40 in 130 people Willingness to participate in research studies. Sampling method was census study. Statistical significance was evaluated by using SPSS Software.

III. RESULTS

Figure I shows the abundance patients in their lodgin that and in Fig II shows clinically signs and symptoms in patients. Also levels of Hb electrophoresis strip method were respectively (MCV<80fl), (MCH<27pg) and (HCT<40), the mean and standard deviation of Hb (mg/ml) were 9.2 and 1.24, MCV(fl) were 87.24 and 0.10 and MCH (pg/cell) were 20.98 and 3.63 and Hb A2 were 2.93 and 0.50 and HCT were 32.30 and 5.43 for both gender.

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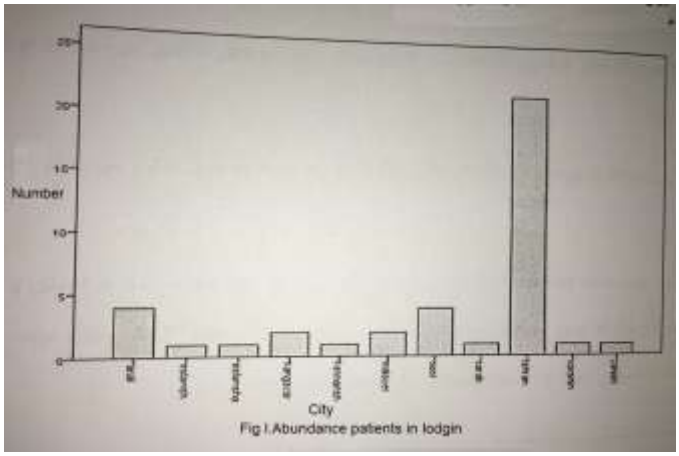

Fig I. Abundance patients in their lodgin

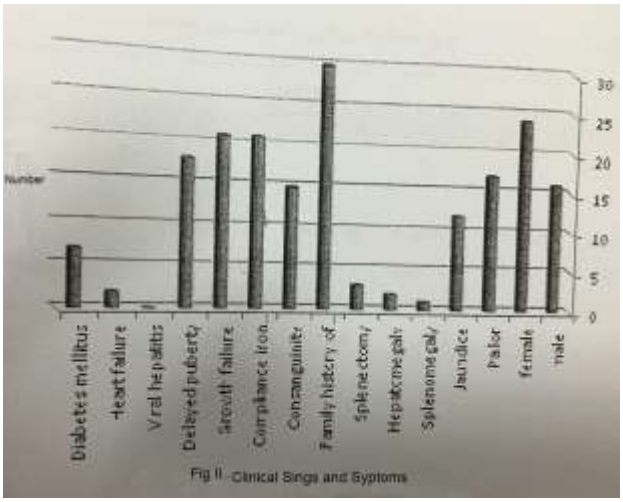

Fig II.Clinical sings and syptoms

V. Discussion

Our results showed that mean and standard deviation of Hb(mg/ml) were 9.2 and 1.24, MCV(fl) were 87.24 and 0.10 and MCH (pg/cell) were 20.98 and 3.63 and Hb A2 were 2.93 and 0.50 and HCT were 32.30 and 5.43 for both sexes.

Also Iron toxicity is a major determinant of disease prognosis in b-thalassemic patients [22]. Although MRI is the gold standard in assessing early iron accumulation in the myocardium, echo is more widely available and cheaper. Monitoring ejection fraction can be useful, but its value is limited by the masking of ventricular dysfunction via the basal high cardiac output seen in chronic anaemia, and its late occurrence in the disease process [23]. Even modern contractile measures, such as tissue Doppler imaging, correlate poorly with cardiac iron. In a TDI study conducted by Silvilairat et *al* [24], diastolic dysfunction was completely absent in patients who had ferritin levels below 2500 ng/ml. This observation was consistent with our results. Furthermore, data by Kremastinos [25] illustrated diastolic abnormalities only after the onset of cardiac failure. Margi et *al* [26] have suggested strain indices as potential markers of early myocardial dysfunction in young asymptomatic patients, who nevertheless had myocardial iron as suggested by the reduced T2* values on MRI. Again in that study all conventional echo measurements failed to illustrate any significance between the patient and the healthy control groups. In the other hand, left atrial volumetric assessment employing RT3D seems to offer an alternative approach as it can provide useful clinical information in early disease states, regardless of the left ventricular ejection fraction [27]. In our study, the impaired AAEF reflects early dysfunction of the left atrium, which could be attributed to the fact that the LA is formed by thin walls that are earlier affected as compared to the left ventricular wall. The early impairment of left atrial function in thalassemia major has also been implicated by studies in asymptomatic thalassemic individuals employing biochemical parameters such as atrial natriuretic peptides [28-29]. In addition to that, Trikas et *al*. have illustrated that LAEF was reduced in well-preserved thalassemic patients, a finding that was verified by our results. According to the same authors, this reduction was associated with reduced exercise capacity [30].

IV. Conclusion

We have shown that reveres slot blot technique or strip can be determine accurately and in less time, without need for sophisticated laboratory equipment the beta-Thalassemia mutations and patients.

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