CODEN [USA]: IAJPBB ISSN: 2349-7750

INDO AMERICAN JOURNAL OF
PHARMACEUTICAL SCIENCES

http://doi.org/10.5281/zenodo.1098160

Available online at: http://www.iajps.com Research Article

A RETROSPECTIVE STUDY ON THALASSEMIA IN SOUTH INDIAN TERTIARY CARE TEACHING HOSPITAL

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Abstract:

Thalassemia syndromes are a heterogeneous group of disorders caused by inherited mutations that decrease the synthesis of adult haemoglobin, Hb-A [alpha2 beta2]. Depending on whether the genetic defect or deletion lies in transmission of alpha or beta globin chain genes, thalassemia are classified in to Alpha and Beta Thalassemia. ***Materials and methods:*** *A Retrospective study was carried out in RIMS Kadapa for two months. Patient demographic details, type of thalassemia, prevalence of thalassemia, Hb levels, known & new cases were collected.* ***Results:*** *A total of 139 cases were diagnosed as having thalassemia in between February 2017 – March 2017. Among which 55 cases were seen in between 11-15 years followed by 6-10 years [38cases] later 0-5 years [38 cases] , 16-20 years [2 cases] 31-35 years[1case]. Highest number of cases was observed in males [91 cases] than females [48]. In our study we couldn't identify type of thalassemia in 122 cases as it was not written in the case sheets.* ***Conclusion:*** *There is a urgent need for making the people aware of this threatening disease Thalassemia, as this disease increasing day by day in the population silently. Health education programmers in the society may reduce the burden of the disease. Moreover, the complications of Thalassemia are one of the major problem on which one has to focus. Diagnosis of Thalassemia, treatment in the early phase may reduce the economic burden and complications in the society.*

Key words: *Alpha thalassemia, Beta thalassemia, RBC destruction, Anemia.*

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Please cite this article in press as J. Lavanya et al., ***A Retrospective Study on Thalassemia in South Indian Tertiary Care Teaching Hospital****, Indo Am. J. P. Sci, 2017; 4(12).*

INTRODUCTION:

The Thalassemia syndromes are a heterogeneous group of disorders caused by inherited mutations that decrease the synthesis of adult haemoglobin, HbA [alpha2 beta2][**1**]. It is a single – gene disorder that is passed from parents to child by what it is called an autosomal recessive pattern of inheritance. It prevents the body from synthesis of sufficient quantity of high quality of blood. Normally an individual inherits two beta globin genes located one each on two chromosomes 11, and two alpha globin genes one each on two chromosome 16 from each parent. Depending on whether the genetic defect or deletion lies in transmission of alpha or beta globin chain genes, thalassemia are classified in to Alpha and Beta Thalassemia. Thus, patients with alpha thalassemia have structurally normal alpha globin chains but their production is impaired. Similarly, in Beta thalassemia, beta globin chains are structurally normal but their production is decreased. Each of the two main types of thalassemias may occur as major or minor. The former is generally assymptomatic while the latter is a severe congenital haemolytic anemia **[2]**. The pathophysiology includes, during pregnancy, the blood of the foetus contains a special kind of haemoglobin, called foetal haemoglobin [HbF], made up of one pair of beta chains and one pair of Álpha-chains [gamma 2 Álpha 2]. This haemoglobin carries out the same function of transporting oxygen around the body that normal haemoglobin performs in older children and adults. After birth, foetal haemoglobin normally continues to function for the first six months of life, when it is gradually replaced with adult haemoglobin [HbA], made up of two alpha-chains and two beta-chains. In thalassaemia major, however, no,beta-chains are produced - or only a very small amount - preventing the synthesis of normal adult haemoglobin and severely damaging the red blood cells' capacity to transport oxygen. The child's body reacts by continuing to make foetal haemoglobin. However, it cannot make a sufficient quantity to meet the needs of the child's growing body and replace the oxygen transporting functions of adult haemoglobin alpha 2 beta 2. Excess alpha chains interfere with the body production of RBC, reducing production by up to 95%.[**3**] Symptoms includes yellowish colour of skin, eyes, pale, fatigue, tachycardia, weak, lethargy, irritable. Complications are Osteoporosis, Osteopenia, splenomegaly, infections, heart failure, endocrine problems like hypothyroidism, hypogonadism, growth failure, Diabetes.**[4]** Treatment involves blood transfusion includes only 250 ml of packed RBC and in order to maintain hyperplasic marrow, folic acid supplement is given. Prevention and treatment of iron overload is done by chelation therapy with desferoxamine and Bone marrow transplantation from HLA-matched donar that provide stem cells which can form normal haemoglobin.**[3,5]**

METHODOLOGY:

The present study was a retrospective study carried out in the General medicine, ENT, Paediatrics, Orthopaedic and Surgery departments medical records in Medical record department of Rajiv Gandhi Institute of Medical Sciences [RIMS] general hospital which is a 750 bedded hospital, Cuddapah, south-Indian tertiary care teaching hospital of Andhra Pradesh, India for a period of 2 months [01 February 2017 to 31 march 2017].

A total of 4200 medical records were reviewed in our study. Among those, 139 medical records regards to thalassemia which were included for conducting this study of all age groups and both genders with past and present Thalassemia disease were included where as thalassemia co-morbid conditions were excluded for conducting the study. All the records were checked by the clinical pharmacist and those who met the study criteria were included. Data required for the present study was acquired from the chart review.

Demographic details like [age and sex], type of Thalassemia, Prevalence of Thalassemia in different blood groups, known and new cases of Thalassemia, hemoglobin [Hb] levels, type of treatment received, amount of blood transfused were collected. Clinical data was studied and analyzed. Data was analyzed by descriptive statics.

We observed 122 cases of Alpha Thalassemia, 9 Major Beta Thalassemia and 8 Beta Thalassemia which were represented in the below table no. 04.

RESULTS:

A total of 139 medical records were enrolled in the study. Data was collected based on the inclusion and exclusion criteria. Demographic data and clinical data was collected and analyzed.

Distribution of thalassemia medical records based upon the age:

A total of 139 medical records were diagnosed as having thalassemia in between the period February 2017 – March 2017. Among those, 11-15 years [39.57%] followed by 6-10 years [30.94%] later on 1-5 years [27.34%], 16-20 years [1.44%] and above 20 year [0.725%]. We found that average age of the patients were 9.123 [±4.507]. It is represented in the below table 1.

Table 1: Distribution of thalassemia medical records based upon the age and gender

Age	Female	Male	Percentage [%]
1-5	15	23	27.34
6-10	13	30	30.94
11-15	19	36	39.57
16-20	1	1	1.44
Above 20	0	1	0.72

Distribution of thalassemia medical records based upon the Gender:
Among 139 records we found 48 were females [34.53%] and 91 were males [65.46%]. Majority of males is higher when compared to females as shown in table 2.

Table 2: Distribution of thalassemia medical records based upon the Gender

Gender	No of patients [n=139]	Percentage [%]
Female	48	34.53
Male	91	65.46

Distribution of thalassemia medical records based on the weight:
Among 139 records, we observed that majority of the patient's weight is in between 11-20 kgs followed by 21-30kgs later on 0-10kgs, 31-40kgs, 41-50kgs and 51-60kgs. We found that the average weight of the patients along with standard deviation was 21.087[±9.012]. It is represented in the table 3.

Table 3: Distribution of thalassemia medical records based on the weight

Weight	No of patients
0-10	12
11-20	60
21-30	52
31-40	11
41-50	2
51-60	1

Distribution of thalassemia medical records based upon the Type of Thalassemia:
In our study we could not able to identify type of thalassemia in 122 cases as it was not mentioned in case sheets, we found 9 thalassemia cases and 8 major cases as represented in the Table 3.

Table 4: Distribution of thalassemia medical records based upon the Type of thalassemia

Age [years]	Beta-T Total	Major Beta- T	Alpha-T	Grand Total
1-5	4	2	32	38
6-10	4	4	35	43
11-15	1	1	53	55
16-20	0	1	1	2
31-36	0	0	1	1
Total	9	8	122	139

Distribution of thalassemia medical records based on the history:
69.07% records were newly diagnosed whereas 30.93% records were known case of thalassemia. It is represented in table .4.

Table 5: Distribution of thalassemia medical records based on the history

Past History	Female	Male	Grand Total	Percentage
Known	12	31	43	30.93
New	36	60	96	69.07

Distribution of thalassemia medical records based on the Haemoglobin level:
The hemoglobin level was 3-4.9 in 07.93%, 5-6.9 in 40.28% and 7-8.9 in 47.48% the remaining above 8.9 in 04.31% was as shown in the table 1.5. Majority of the patients was seen with Hb level 7-8.9 in 66 records followed by 5-6.9 in 56 records and 3-4.9 in 11 records. We found that average levels of Hb were 6.718 [±1.388].

Table 6: Distribution of thalassemia medical records based on the haemoglobin level

Haemoglobin levels [gm/dl]	No. of medical records	Percentage [%]
Less than 3	0	0
03 - 4.9	11	07.91
05 - 6.9	56	40.28
07 - 8.9	66	47.48
9 & above	6	04.31
Total	139	100

DISCUSSION:

Thalassemia is the genetic disorder occur primarily due to defective formation of globin chain of the hemoglobin moiety of the RBC. It is a specific type of blood disease which results in consequences of excessive destruction of red blood cells which in turn leads to anemia. In this, RBC breakdown occur at an early stage due to abnormal globin chain unable to protect RBC in oxidative stress. Ultimately, resulting in destruction of RBC. In thalassemia the rate of destruction of RBC is so rapid that it exceeds the liver capacity to metabolize the excess billirubin. Thalassemia is a major health problem, and approximately 1 in 14 of the population is carriers for one of the sub types.

As it is a retrospective study, our study focuses on the prevalence of Thalassemia and their types in the population. In our study 139 thalassemia medical records were enrolled in that 48 were females and 91 were males. Our study reveals the actual rate of the population suffering from Thalassemia is male and the incidence of male population is higher. In this study, we categorized the patients according to their age groups and found that the majority of the patients suffering from Thalassemia are in the 11-15 years age group. **CK Li, CW Luk et al.,** also reported that majority of patients with thalassemia are in age group of 11-20 years. In our study population the patients came with known history of Thalassemia is more when compared to the new cases. Health education is important in both patients as there is a chance of getting Thalassemia to their children. It requires proper disease and treatment education and adequate knowledge regarding their lifestyle modifications to overcome the situation in the family or community. In our study, we found majority of the patients are with hb levels lower than 9 i.e., 95.69%. **Abdolreza Rajaeefard et al.,** also concluded that majority of the patients are with below 9 Hb levels. We distinguished the thalassemia medical records based on the type of Thalassemia and found that patients suffering from beta thalassemia type are more. Educating the person about types of Thalassemia is much more important as this type also leads to anemia. Complications of Thalassemia and blood transfusion compatibilities must be understood by every individual.

CONCLUSION:

There is an urgent need for making the people aware of this threatening disease Thalassemia, as this disease is mounting day by day in the population silently. Health education programmers in the society may diminish the burden of the disease. Moreover, the complications of Thalassaemia are one of the foremost problem on which one has to be focus on diagnosis of

Thalassemia, treatment in the early phase may reduce the economic burden and complications in the society. We observed Thalassemia majorly in the pediatrics. Hence, it is important to provide patient counseling by the clinical pharmacist to the parents and the individuals for better health care.

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