Prevalence of thalassemia in and around Pune District, Maharashtra

1-Dra Kalpana B Rathod, 2-Dr Deepak G Kulkarni, 3-Dr Zeeshan F Hashmi, 4-Dr. Shilla C Puranik

1Assistant Professor, 2Associate Professor, 3SR, 4Professor and HOD.
Department of Pathology, BJ Government Medical College and SGH Pune, Maharashtra 411001
Corresponding Author Dr Kalpana B Rathod

Abstract
Thalassemia are heterogeneous group of disorders resulting from abnormalities of globin chain synthesis. Which in turn result in abnormal haemoglobin synthesis.
The present study includes total number of thalassemia patients in and around Pune District, Maharashtra during the period from 1st December 2015 to 30th November 2016. This original article showed a prevalence of thalassemia to be around 6.2% in total number of admitted patients in paediatric ward.
Key Words – Thalassemia, Prevalence, Consanguineous

Introduction
India is a country with many religions which are divided into many castes. There are around 3000 castes and 25,000 sub castes in India. Consanguineous marriage remains the choice of an estimated 10.4% of the global population (1). There are more consanguineous marriages taking place in India and the premarital checkup is not under process in India, which leads to many genetic disorders. Many Diseases are passed on from parents to their children’s. Thalassemia is one of them. Thalassemia is present almost in every community. Thalassemia is caused by a genetic inability to make normal amounts of haemoglobin (2). The thalassemia’s are heterogeneous group of disorders of haemoglobin synthesis resulting from the reduced rate of synthesis of one or more globin chains of haemoglobin. Decreased synthesis of α chain produces α thalassemia, while decreased synthesis of β chains produce β thalassemia. The thalassemia are among the most common genetic disorders worldwide, occurring more frequently in Mediterranean region, the Indian subcontinent, Southern Asia, and West Africa. (3)
If there is a reduced output of α chains, any excess of γ chains or β chains produced will give rise to molecules with the formula γ4 (Hb Bart’s) and β4 (Hb H), respectively. Hence the α thalassemia’s are usually associated with the presence of Hb Bart’s in infancy and Hb H in adult life. (4)
Thalassemia is a major health problem, placing an immeasurable emotional, psychological and financial burden (5,6) Screening can be done for thalassemia in four different ways. They are prenatal, new born, premarital and random total population screening. Thalassemia minor screening would help to reduce the thalassemia major cases. Screening can be done using different methods. Initially, a complete haemogram is done in which the MCV less than 80fl would be further screened using Naked Eye Single Tube Red Cell Osmotic Fragility Test (NESTROFT). The person who shows positivity for NESTROFT would be checked for Hba2 level using HPLC or Hb electrophoresis.
Mutations can be checked by PCR and automated sequencer.(7)

**Materials and methods**

Present study was carried out in the department of Pathology, BJ Government Medical College and SGH Pune from 1 Dec 2015 to 30 Nov 2016. The study included all thalassemic patients from paediatric ward. They are from Pune district and adjoining areas. These are the patients who come from these different places for medical treatment, wherein we have prepared a separate thalassemia ward and patients are admitted in specified ward for necessary treatment in form of blood transfusion and drugs. The 276 total number of patients were found during above period.

**Result**

In the present study 276 patients are detected in above period. The distribution of patients are shown in the following tables.

Table No 1 Age distribution

<table>
<thead>
<tr>
<th>Sex</th>
<th>Total No.</th>
<th>Minimum Age</th>
<th>Maximum Age</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>139</td>
<td>8 months</td>
<td>11 years</td>
</tr>
<tr>
<td>Female</td>
<td>137</td>
<td>5 months</td>
<td>14 years</td>
</tr>
</tbody>
</table>

Table No 1 shows total number of thalassemia patients in paediatric ward with age distribution.

Table No.2 Haemoglobin

<table>
<thead>
<tr>
<th>Sex</th>
<th>Average gm %</th>
<th>Minimum gm %</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>7.37</td>
<td>5.5</td>
</tr>
<tr>
<td>Female</td>
<td>7.17</td>
<td>4</td>
</tr>
</tbody>
</table>

Table No 2 shows average and minimum haemoglobin in the cases studied.

Diagram 1- Showing thalassemia patients out of total indoor patients in paediatric ward.

4442-Total patients in paediatric ward - indoor
276-Total thalassemia patient, Prevalence of thalassemia 6.2%
Maximum indoor patients presented with anaemia, few showed thrombotic complications and very few presented with bony deformities associated with osteoporosis.

**Discussion**

Ten percent of the total world thalassemics are born in India every year (8). It has been estimated that the prevalence of pathological hemoglobinopathies in India is 1.2/1,000 live births (9), and with approximately 27 million births per year (10). This would suggest the annual birth of 32,400 babies with a serious haemoglobin disorder.

The first case of Thalassemia, described in a non-Mediterranean person, was from India. Subsequently, cases of thalassemia were documented from all parts of India (11).

Thalassemia treatment is costly. To get an ideal treatment for one thalassemic child it costs around Rs. 1,25,000/annum. Hence for 50,000 children the cost would be nearly Rs.620 crores. This staggering cost is beyond the reach of our country, India; moreover, this cost is expected to increase due to additional children being born (12).

Thalassemias are a major health problem, and approximately 1 in 14 of the population are carriers for one of the sub types. Over the past three decades, regular blood transfusions and iron chelation have dramatically improved the quality of life and transformed thalassemia from a rapidly fatal disease in early childhood to a chronic disease compatible with prolonged life (13,14). Today life expectancy varies between 25-55 years, depending on the compliance with medical treatment (15,16).

Despite increased life expectancy, complications keep arising.

Health education is an important component of the preventive genetic programs. This requires proper health education and adequate sensitization to the individual, family or community to accept these preventive remedial measures. High cost of treatment, repeated blood transfusion and chelation therapy, and economic burden on family resources, all suggest that prevention is better than cure. Thus a joint venture of antenatal and inductive screening seems to be the most fruitful strategy for beta thalassemia in India. With improving environmental and socio-economic conditions, better public health care and medical facilities and better nutrition, children suffering from thalassemia and hemoglobinopathies can be better managed and rehabilitated in India (17-20).

**Prevention of thalassaemia**

The need for prevention of thalassaemia is obvious due to high frequency of the condition, the great expense and difficulties in providing optimal treatment for patients, and the innumerable fatalities from untreated β-thalassaemia. Prevention would not only be a good public health practice, as envisioned in Alma Ata declaration, but it would also be cost-effective, as the ratio of the cost of treatment to prevention is 4:1, as shown in a study from Israel.

It would help tremendously in reducing the burden of the disease for patients, families and the health services. The strongest argument for prevention is that it would ensure the best possible care for the affected, by curbing the increase in their number.

The chief elements of a control programme were developed in 1970s by a team of experts at the World Health Organization, led by Dr Bernadette Modell. These are (i) Political and financial support, (ii) Improving curative services; (iii) Prenatal diagnosis in couples who have given birth to an affected child, as well as those identified to be at risk, (iv) Prospective antenatal screening, (v) Community carrier screening, (vi) Counselling and prenatal diagnosis, and (vii) Network of centres, and National/Regional working groups.
Outcome of patients in the form of blood transfusion and drug therapy is much better and life pattern of patient is more comfortable. More emphasis should be given on health education of the society the result of consanguineous marriages and how to prevent the disease from occurring in the community.

References