Original Article

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Screening of β-Thallasemia in Tribal Population of Hingoli District

Raviraj Naik*, Sarita Dakhure*

Author Affiliation: *Assistant Professor, Department of Pathology, Indian Institute of Medical, Science & Research, Aurangabad-Jalna Road, Warudi TQ, Badnapur Distt, Jalna-431202 (Maharashtra).

Abstract

The cross sectional study was performed to find out the incidence of β-Thalassemia trait in tribal population of Hingoli district; Maharashtra from July 2014 to May 2016. The study was conducted at IIMSR,Medical College,Jalna. In this study we screened 1075 tribal subjects comprising adult men & women as wellas children. Whole blood Samples were collected in EDTA bulb for Naked Eye Single Tube Red cell Osmotic Fragility Test (NESTROFT). The screening of â thalassemia trait was done on NESTROFT with 0.36% freshly prepared saline. Out of 1475 tribal subjects the NESTROFT was positive for 111 subjects which indicated very high chances of subjects having β-Thalassemia trait. With this rate of β thalassemia trait, urges necessity in studying beta thalassemia carrier status in the child bearing group, as a primary step to prevent the birth of beta thalassemia major.

Keywords: Tribal; β-Thalassemia Trait; NESRTROFT.

Indroduction

β- thalassemia is one of the most common single gene disorders in India with an overall prevalence of 3-4% [1]. In certain communities like Sindhis, Muslims, Cutchi Bhanushalis, and some tribal groups, the prevalence of β thalassemia carriers varies between 8 and 10% or more [2,3].

Reportedly, there are about 240 million carriers of â-thalassemia worldwide and in India alone the number is approximately 30 million with a mean prevalence of 3.3% [4,5]. It has been estimated that about 10 000- 12 000 children with â thalassemia major are born every year in India. These figures might be underestimated. As the frequency of thalassemia is increased by the consanguinity and endogamous mating, it may be assumed that the Sindhi communities in India are facing the problem at large scale. Three classes of β-thalassemia have long been recognized clinically, β-thalassemia major, intermedia and minor[6]**.** β- Homozygous state presents with variable degree of anemia from early childhood and are generally transfusion dependent, a condition clinically known as thalassemia major. β-heterozygous cases (thalassemic minor) are almost asymptomatic with normal or slightly reduced levels of hemoglobin. However an intermediate condition which may have either heterozygous or homozygous pattern of inheritance, requires minimal or no blood transfusion and has milder clinical course than thalassemic major but is severe enough as compared to thalassemic minor. It manifests generally after two years of age and does not require regular transfusion therapy [7,8].

There is growing concern that thalassemia may become a very serious problem in the next 50 years, one that will burden the World's blood bank supplies and the health system in general. Therefore emphasis has shifted from treatment to prevention of birth of such children in future [9]. The most effective approach to reduce the burden on the society and to reduce the disease incidence is through

Reprint Request: Raviraj Rajan Naik, IB 66, OM Bungalow, Behind Nath Seeds, Paithan Road, Aurangabad 431005. Maharashtra.
E-mail: raviraj_40@yahoo.com

implementation of a carrier screening programme, offering genetic counseling, prenatal diagnosis and selective termination of affected fetus [10]. Need for prevention of thalassemia is obvious due to high frequency of the condition, the great expenses and difficulties in providing optimal treatment for patient. Prevention would not only be a good public health practice, but would be cost effective, as the ratio of treatment to prevention is 4:1 as shown in the study from Israel [11].

Tribal population in India is not a homogenous group. Further, because of their isolated existence and endogamy over centuries, different tribal populations have distinctive genetic identities. With industrialization and availability of jobs in different areas of India many of these tribal populations have migrated from their homelands to cities in search of jobs. Although these migrations are relatively in small proportions (5-10%) but the absolute number of tribal persons living in big cities and industrialized areas of the country is substantial [12].

In context of the achievement of education and exposure to developing world, the Indian tribes are far from using the 'freedom in choice' which basically relies on the dissemination of accuracy in information and sharing that at social and familial level. Delicate and proper understanding of the very sensitive psycho-social determinants and aspiration of the tribal societies need to be considered before implementing any counselling effort pertaining to haemoglobinopathies in India.[13]

Tribal groups in Maharashtra have shown a prevalence of β-thalassemia trait of 1.6 to 5.6 % [14].

Tribal population comprises 35-40% of total hingoli population.Common tribes in hingoli district are andh, bhill and thoti mostly dwelling in small villages of district. Investigations like haemoglobin electrophoresis & HPLC are helpful in diagnosing beta thalassemia . However, these investigations are either expensive or time-consuming or cumbersome and often require sophisticated equipment. Hence, cannot be used as effective tools for population screening.

For screening purposes, a test which is inexpensive, requires a small amount of blood, does not require sophisticated equipment and can be applied on the population as a whole is preferred. These requirements are met by a modified osmotic fragility test "NESTROFT" (Naked Eye Single Tube Red Cell Osmotic Fragility Test), a test first described by Kattamis et al [15].

Aims & Objectives

1. To study the prevalence of β– thalassemia trait in tribal people of hingoli district.
2. To make tribal people aware of prevention and management of β- thalassemia in order to decrease the burden of morbidity and mortality associated with the disease.

Material & Methods

Study Design

Cross sectional study.

Study Period

July 2014 to May 2016.

Ethical Approval

The study was approved by the IIMSR,Jalna Institutional Ethical Committee and due permission was taken from civil surgeon office at Civil hospital hingoli.

Inclusion Criteria

All the tribal subjects in the age range of 3 to 35 years were included in this study. This study aimed at targeting children and young population.

Site of Sample Collection

OPD of Civil hospital at hingoli city & Rural hospitals at kalamnuri , salegaon , aundha which are talukas of Hingoli district. Sample were collected in four phases in the form of camps organized at civil & rural hospitals of kalamnuri,aundha & salegaon taluka by Dr.Raviraj Naik and Dr. Sarita Dakhure.

Storage of Sample

Sample collected were stored in 3ml plastic cuvetts in freezer compartment of refrigerator.

Site of Sample Study

Biochemistry laboratory IIMSR,Jalna .

Method

3 ml of blood sample was collected in EDTA bulb, and all samples were screened for beta thalassemia

trait by using NESTROFT with 0.36% buffered saline solution. 2 ml of the 0.36% buffered saline solution was taken in one tube (10 cm x 1 cm diameter) and 2 ml distilled water was taken in another tube. A drop of blood was added to each tube and they were left undisturbed for 1/2 an hour at room temperature. Both the tubes were then shaken and held in NESTROFT test kit stand having white background with thin black line drawn over it. The line was clearly visible through the contents of the tube containing distilled water. If the line was similarly visible through the contents of the tube with the buffered saline, the test was considered negative. If the line was not clearly visible, the test was considered positive. A positive test indicates lowered red cell osmotic fragility, suggestive of thalassemia trait, and confirmed by Hb A2 level >3.5% performed by HPLC.

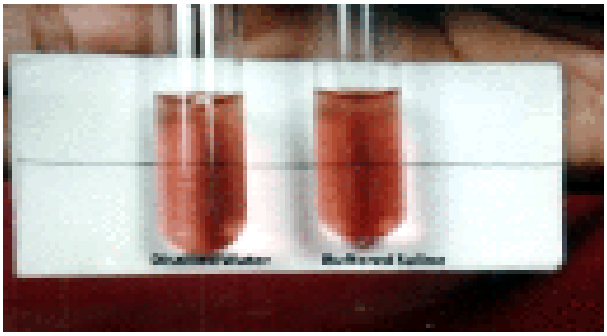

Fig. 1: Photograph showing negative NESTROFT

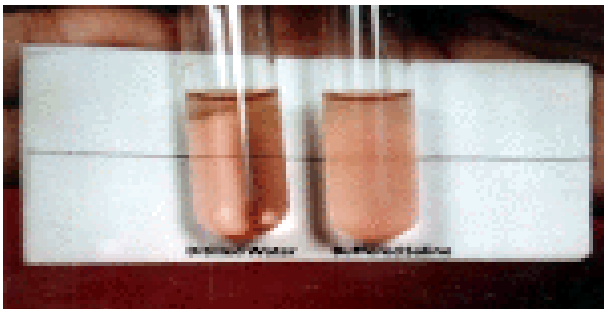

Fig. 2: Photograph showing positive NESTROFT

The tubes were left undisturbed for 3 hours. At the end of 3 hours, the DW tube was seen to be homogeneously pink with no sediments. In the BS tube the negative test showed similar findings as DW tube where as in a positive case, a clear supernatant and a sediment at bottom was observed [16].

Observation & Results

Blood samples of 1475 tribal people including men,women & children were taken for the study. The samples were subjected for NESTROFT test as they were available. After analyzing the data it was found that out of 1475 tribal people, 111 showed NESTROFT positive. So out of 1475 tribal subjects 47 men, 51 women and 13 children(8 boys & 5 girls) gave the NESTROFT test positive.

Table 1:

City/Taluka	Total Tribals Screened	Total tribals +ve for Nestroft test
Hingoli City	215	18
Kalamnuri taluka	566	37
Aundha taluka	445	29
Salegaon taluka	249	27
	1475	**111**

None of the tribal subjects which were positive for nestroft test were suffering from any clinical features of thalassemia; so they were labeled as silent carriers. Only 5 subjects were aware of the disease called as thalassemia and none of them were knowing about common occurrence of thalassemia in tribal population. Only 9 subjects among total screened were aware of the fact that consangious marriage should be avoided. All tribals arrived at camps at all 4 hospitals were councilled in simple and local language regarding prevention and management of thalassemia and also were advised all doand don't do in social life in order to decrease the burden of suffering from â thalassemia diasease in tribal society.

Those tribal subjects who gave nestroft test positive may be labeled as having β-thalassemia trait but nestroft test does not confirm the disease. Ideally subjects giving netsroft test positive should be further investigated by running their sample in Hb electrophoresis or HPLC which is best in this regard. Unfortunately we were not equipped with HPLC at our institute so samples were not further processed.We felt no need of sending the sample to other institute as none of nestroft positive subjects were symptomatic and severe anemic (Hb<7gm%).

Discussion

This study revealed that almost 7.52% of tribal population in hingoli district was suffering from β-thalassemia trait. Similar findings were reported by Sukumari et al [2] and Mehta et al [3]. However previous studies revealed that the tribal groups of India have high risk of beta-thalassemia, the prevalence of carrier status in some being as high as 17% [17]. Agarwal et al. [18] stated that the majority of the beta thalassemia carriers were of Uttar Pradesh origin. But according to Verma et al. [19], the majority of β-thalassemia carriers in India were migrants from Pakistan and their pattern of mutations differed from

the rest.

On the basis of earlier reports published since last 20 years, it is clear that several tribal groups of India have been identified as high-risk groups for thalassemia and other haemoglobinopathies. It causes high degree of morbidly and mortality among them [20]. In India, with about 4635 ethniccommunities five common and 12 rare mutations have already been reported [21,22]. Previous studies have given some probable causes of such high frequency of the disease among the tribal people. Migration of tribal groups from higher risk zone may be one of the causes of having high prevalence of [23,24] Similar haplotype for tribal groups from different parts of India may be consistent with the hypothesis of the Unicentric origin of the mutation in the globin chain as well as Unicentric origin of the tribal population.Due to the practice of non-random mating pattern for a long time, some particular mutations are restricted to some specific groups [25]. High inbreeding rate due to consanguineous practices of marriage make this situation more complex, because consanguinity is an important issue to spread the disease [26]. In addition to this, lack of proper medical facilities, natural barriers like forest, ecological niches etc., poverty, illiteracy, poor sanitation, lack of safe drinking water, faith in traditional beliefs and taboos [27] have further compounded the complexity [28].

Despite of considerable advancement in management strategies of thalassemia in India, the problem still remains for tribal and isolated populations. Because implementation of technological advances to the realities of health care in developing countries is a challenge. The strategy that has been taken in India is sometimes problematic to implement. Thus the problems related to the tribal groups claim a special intervention strategy for prevention and control of thalassemia, which may be more feasible for the Indian tribes. A strategy model for controlling thalassemia among the tribes of India may include treatment and prevention. The only curative treatment available is bonemarrow transplantation and iron chelation, which is highly expensive and not easily affordable by a tribal family. Prevention and control of new thalassemic baby is, therefore, more important to reduce the prevalence of these diseases.Prevention can be done through increasing the awareness and testing at a mass level. But lack of awareness and indifferent attitude towards thalassemia is very common among the tribal people. They are ignorant about the medical, social and financial burden of the disease. Thus in this prevention program priorities should be given on the public awareness, which can be done through community education, awareness camp, awareness at school level and motivation of high risk group. For these, schools, college and different government sectors may play major role with active involvement of media. In remote areas where even the media is not accessible, NGOs can take the responsibilities. An ideal screening programme of thalassemia trait for tribal woulde consist of definitive strategies such as ; extended family screening (i.e. the testing of the relatives of thalassemia patients), as the firstdegree relatives of a thalassemic patient have “14% higher risk of having an affected child compared to the general population”. Second level of testing is the carrier screening of unmarried girls and boys of the tribal communities. Next strategy that can be adopted is the genetic counseling of married couple before pregnancy.

Another strategy for screening the target population may be the testing of pregnant women attending hospital / healthcare unit, after which the husband is asked to be tested if the wife is found to be a carrier. The later one is a cost effective strategy as it reduces the screening cost by 50%. This test should be mandate for each and every pregnant woman and should be free of cost for them. And if the result of the test is positive, then the only way is the selective termination of affected fetuses. Apart from this, it may be noted that only awareness is not enough for tribal communities. Constant monitoring is essential through community participation which is possible only when health setup comprising of PHC & Rural Hospital are fully bestowed with facilities required for investing haemoglobinopathies such as thallasemias.

Conclusion

1. Out of 1475 tribal people screened, 111 showed NESTROFT positive and may be labeled as having â-thalassemi trait.
2. Tribal people should be made aware of different hemoglobinpathies common in tribal population and should be councilled properly to give up the dangerous tradition of consangious marriages.
3. Much further study needs to be undertaken in order to investigate for other hemoglobinpathies which are common in tribal population.

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