ORIGINAL ARTICLE

# Prevalence of ß-Thalassaemia in African Descendent Lyari Population

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## ABSTRACT

**Objective:** To assess the prevalence of beta thalessemia Trait in the African descendant Population, of Layari Karachi Pakistan.

**Study Design:** Descriptive cross-section study

**Place & Duration:** Samples collected at Lyari General Hospital, Karachi & Tested at Dow Institute of Haematology . DUHS. Karachi, from October 2012 March 2013.

**Material & Methods:** We studied 200 consecutive subjects of Makrani cast of both genders, who visited Lyari General Hospital during the study period. The demographic data was entered on a proforma. CBC, peripheral smear and haemoglobin electrophoresis was performed on all samples.

**Result:** The male to female ratio of the study population was found to be 0.59:1. 5 patients of ß-thalassaemia major and 27 patients of ß- thalassaemia trait were detected on electrophoresis with overall frequency of 16% , 2.5% for major and 13.5% for trait in our study population. The overall percentage of family marriages in the study group was 59%. In thalassaemic trait group it was 77%, while in diseased group 100% of the patients gave history of parental consanguinity.

**Conclusion:** The prevalence of ß-thalassaemia in Makrani cast of Lyari is found to be quite high i.e. 16%,common custom of family inter marriages was also detected which may be one of the cause of high frequency of haemoglobinopathies in that cast.


**Keywords:** Complete Blood Count, Thalassaemia, Anaemia, Haemogram, Electrophoresis.

## INTRODUCTION

Thalassaemia is a genetic disorder in which there is abnormality in haemoglobin tetramer due to some alteration in formation and structure of the globin chains[1]. Thalassaemias are those broad groups of monogenic, recessively inherited disorders in which there is quantitative reduction in globin chain production[2]. There is different sub- types of thalassaemias but the most common is the ß- one[3].

According to World Health Organization 60,000 infants are born every year with this pathology[4] & around 1·5% people in the world carry the mutations for this abnormality[5]. This disorder has spread globally with incidence is increasing alarmingly and it was estimated that around 900,000 infants are expected to be born with this disorder in the next 20 years[6]. Prevalence of thalassaemia in Pakistan is reported in the range of 3-8%[7].

Different studies were performed to elucidate the fact that ß-thalassaemia trait is somewhat protected in malaria endemic areas and because of the natural selection the frequency of this diseases (traits) is more in those areas as compared to the rest of the world[8], furthermore, the custom of consanguineous marriages is also more common in these parts leading to the


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increment of disease burden in these areas[1,9].

More than 1 in 5 people of high-risk ethnic background (from the Middle East, Southern Europe, Indian subcontinent, Central and South East Asia and Africa) may carry a ß-thalassaemia mutation[10] and Lyari is mainly populated with African descendent (Makrani /Sheedi) cast which are included in high risk group. Thalassaemia International Federation statistics shows that from around the world approximately 200000 thalassaemia affected patients are registered for regular treatments patients with and this is not reflective of the true burden of this disease[11], chances are there many patients are not diagnosed because of the financial, economical and other factors and are not even getting the treatment[12].

The above statistics indicate that thalassaemia not only considerably contributes to the economic burden of the country but it is also a major reason of morbidity and mortality.

Our country is a developing country and Lyari in particular is resided by financially deprived inhabitants[13], also, the custom of consanguineous marriages is a norm in those communities, so, there is a dire need to find out the true burden of this disorder in that area. The aim of our study was to evaluate the prevalence of thalassaemia in a high risk Makrani (African descendent) population of Lyari.

## MATERIAL & METHODS:

This cross-sectional study was conducted from October 2012 till March 2013. 200 subjects of Makrani cast of both genders, who came to Lyari General Hospital, Karachi, either as an out patient, attendent to the patient or were admitted there were included in study. EDTA anti-coagulated whole blood sample was collected and evaluated on automated haematology cell counter cell TAC-ß at Dow Institute of Haematology (DDRRL). An 18 parameter CBC haemogram was obtained peripheral smear was prepared within a standardized time limit of 4 hours while Leishman's stain was utilized for staining. Peripheral smear morphology was reported by conventional microscopy. Electrophoresis was performed on all the samples. Study variables were age, gender, Hb%, MCV (Mean corpuscular volume), MCHC (Mean Corpuscular Hemoglobin Concentration), TLC (Total Leucocytes Count), Platelet count, Red cell morphological patterns and electrophoresis results.

## RESULTS:

The male to female ratio of the study population was found to be 0.59:1. 5 patients of ß-thalassaemia major and 27 patients of ß-thalassaemia trait were detected on electrophoresis with over all frequency of 16%, 2.5% for major and 13.5% for trait in our study population (Fig 1). The most common age group for ß-thalassaemia major was 1-10 years (Fig 2) and that for trait was 21-30 years (Fig 3).

The overall percentage of family marriages in the study group was 59%, while in thalassaemic trait group it was 77% and in diseased group 100% of the patients gave history of parental consanguinity (Fig 4). Haemogram indices in diseased and trait subjects are shown in table 1. The most common morphology on peripheral smear was found to be hypochromic, microcytic cells with NRBC's , target cells and poikilocytosis.

**Table-1.** Haemogram indices with ± Standard Deviation in Thalassemia Trait and Dise

| Haemogram Indices | Thalassaemia Trait | Thalassaemia Disease |
|---|---|---|
| Haemoglobin(gm%) | 10.6±2.07 | 5.5±2.3 |
| Red Blood Cell ($10^{12}$/L) | 5.11±0.77 | 2.3±0.75 |
| Haematocrit (%) | 33.8±7.93 | 16.8±8.19 |
| Mean Corpuscular volume (fL) | 69.3±7.2 | 72.6±1.28 |
| Mean Corpuscular Haemogloin (pg) | 21±2.99 | 23.8±5.44 |
| Mean Corpuscular Haemoglobin concentration (g/dL) | 29.9±1.91 | 33±8.12 |

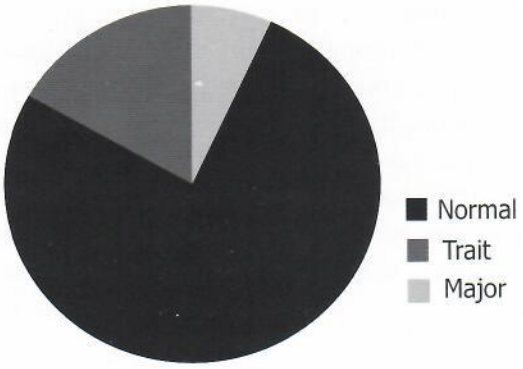


**Fig-1.** Frequency of ß-Thalassaemia trait & Disease in Study Population

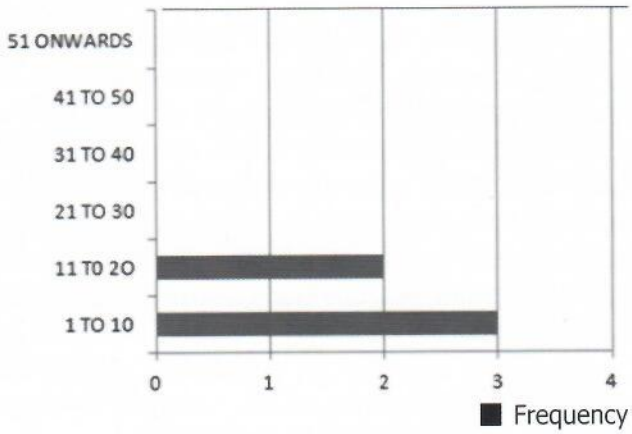


**Fig-2.** Age Group Distribution for *Thalassaemia Major*

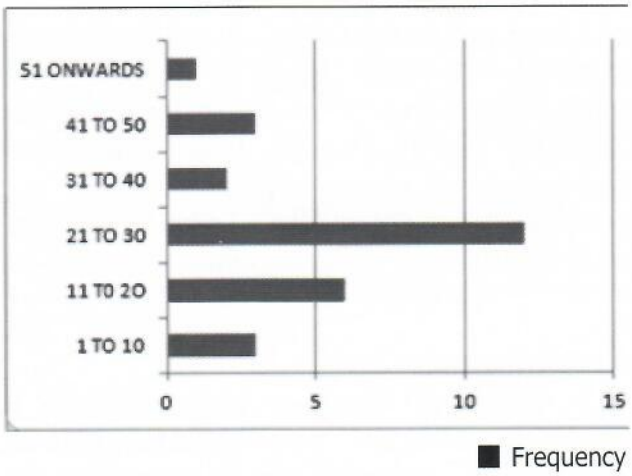


**Fig-3.** Age Group Distribution for Thalassaemia Trait

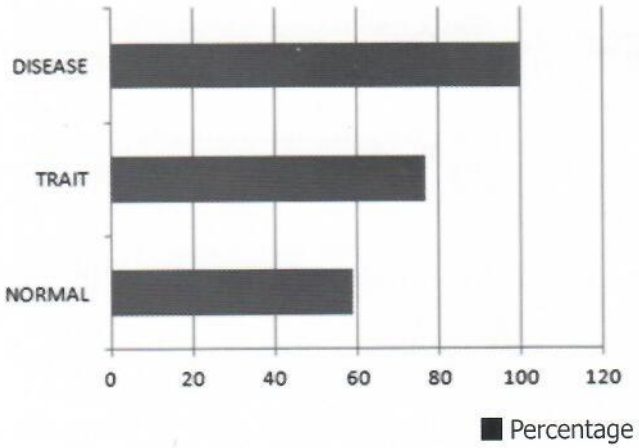


**Fig-4.** Percentage of Family Marriages/Consanguinity

## DISCUSSION:

In Bulletin of World Health Organization, 2008 Public health review was published in which estimated burden of haemoglobinopathies in different countries was mentioned. According to this review there are minimum of following annual births of ß-thalassaemias in different countries as, 1386 in Africa, 341 in America, 9914 in Eastern Mediterrarian, 1019 in Europe, 20420 in south east Asia and 7538 in Western Pacific[14]. High birth rate in certain areas exhibits that haemoglobinopathies are more common in certain ethnic populations[10,14]. Our study was also aimed to estimate the frequency of ß- thalassaemia in one high risk ethnic group and we found 2.5% frequency of ß-thalassaemia major and 13.5% for thalassaemia trait in Makrani population of Lyari.

The Genetic Blood Disorders Survey was done in Oman in 2004 with the aim to determine prevalence of hematological disorders of genetic origin in children under 5 years of age and they found out that ß-thalassaemia trait is present in 2% of those children while, 0.07% of these children had ß-thalassaemia major[15]. These frequencies are much less than the frequencies that we found in our study and this may be due to the fact that we included all age groups in our study while this survey was performed in the children less than 5 years of age. A similar study was conducted in school children (6-15 years) of Jordan and the estimated frequency of ß-thalassaemia minor was 3.04% .

This study found only 1 case out of 1020 (0.001%)[16] of ß-thalassaemia disease again the comparatively lower frequencies can be due to difference in age group inclusion criteria.

A study was performed in Guangdong Province of China in 2003 to find out the spectrum of α and ß-thalassaemia and though increased frequency of ß-thalassaemia was found in the population but 2.54% were positive for ß-thalassaemia trait and 0.26% of the population were having both α and ß-thalassaemia[17]. The frequency of ß-thalassaemia trait is much higher in our study compared to this one, inclusion of high risk population in our study can be one of the major factor for this discordance in results.

An educational and screening programe was conducted in three high schools of Hong Kong, to find out the prevalence of α and ß-thalassaemia. This study found 5% carrier rate for ß-thalassaemia 3.4% were carrier of ß-thalassaemia or HbE, while on the other hand 6 out of 1800(.003%) were carriers for α and ß- thalassaemia[18]. In contrast to our study, in which we included all age groups, this study included only students of high school and the study population was also not the high risk as compared to the ones our study and it may be because of these above mentioned reasons that our study found comparatively high frequency of ß-thalassaemia in our study population.

A study in India using NESTROFT as a screening technique estimated 8.5% prevalence of â-thalassaemia trait in pregnant women of Karnatka (Banlglore)[19], while another study in Nagpur city India using the similar technique for screening concluded the prevalence of 16.81% for ß-thalassaemia in Sindhi population[20]. The estimation of such high prevalence of the trait in the study population is in concordance with the results of our study as only high risk ethnic groups were screened in both studies.

Pre-marital screening in Saudi Arabia found out 3.4% frequency of ß-thalassaemia trait in their population[21], this high frequency calls for pre-marital screening in all high risk populations like study group so that marriages of traits can be prevented. Another Saudi Arabian study which Was conducted to estimate the prevalence of Haemoglobinopathies in different tribes of the country established overall prevalence of 3% for ß-thalassaemia trait, 0.9% for ß-thalassaemia and 43.3% for ß-thalassaemia[22]. But it also concluded that tribal variations also occur for prevalence of these genetic haemoglobinopathies. This tribal variation is also in concordance with the findings of current study in which we estimated high frequency of ß-thalassaemia in our study population which is in contrast to the comparatively lower prevalence in other ethnic groups of our country and this emphasizes the point that this haemoglobinopathy is more common in certain ethnic groups[10,14,22].

Frequency of ß-thalassaemia is estimated to be 1% by a study but this study also concluded that Black persons residing in Comoron are more at risk as compared to the other ethnic backgrounds[23]. Study on ethnic specific rates of Sickle cell anaemia and ß-thalassaemia (1999) concluded that ß-thalassaemia is present in all the ethnic groups in United Kingdom, but, this study also concluded that highest frequency of this disorder is seen in Cyproits[24]. On the other hand National register of surveillance of ß-thalassaemia in 2001 commented that residents of Pakistani origins are now at the highest risk of thalassaemia replacing the Cyproit population[25].

While, as far as Pakistan is concerned no nationwide data involving all the ethnic groups is available but one study done in Northern areas of Pakistan concluded that there is overall prevalence of 5.4% in all ethnic groups of Northern areas while, Pathans have 7.96% and Punjabis have 3.26% of the frequency[26]. This data cannot represent the true picture of the prevalence in our country as it does not involve all ethnic groups and secondly, the most high risk ethnic group of our country i.e. Balochs are not included.

## CONCLUSION:

The prevalence of ß-thalassaemia in Makrani cast of Lyari is found to be quite high i.e. 16%, common custom of family inter marriages was also detected which may be one of the cause of high frequency of haemoglobinopathies in that cast.

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