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Prevalence of thalassemia and sickle cell anaemia carriers among antenatal women– A first study from Telangana population

Padma Gunda[1], Saroja Kondaveeti[1], Mamata Manne[1], Anitha Appam[2], Suman Jain[1,*]

[1]*Thalassemia and Sickle Cell Society, Hyderabad, Telangana, India*
[2]*Government Maternity Hospital, Koti, Hyderabad, India*

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ABSTRACT

Beta thalassemia and sickle cell anaemia (SCA) are highly prevalent and inherited preventable blood disorders that require lifelong blood transfusions and contribute to infant and childhood morbidity and mortality. It is mandate to prevent these disorders to save the children from life long suffering by initiating stringent screening programs. A total of 2478 antenatal women visiting Modern Govt. Maternity hospital (MGMH), Petlaburz, Hyderabad (2000) and Primary Health Care centers of Balanagar (254) and Rajapur (224) in Mahbubnagar district were screened for thalassemia and SCA using Complete blood count followed by High Performance Liquid Chromatography to detect the carrier status. Husbands were screened in case of carrier women and if both the couple were carriers then they were advised for prenatal diagnosis. Out of the 2478 antenatal mothers screened, a carrier frequency of 3.05% and 4.6% was detected for different types of hemoglobinopathies from MGMH and PHC's respectively. Of the different types of thalassemia, beta thalassemia was the predominant one with a frequency of 2.15% from MGMH and 2.9% from PHC's. Three couples were found to be thalassemia carriers and went for prenatal diagnosis. In all the three cases the fetus was found to be normal and they are continuing with the pregnancy. The present study is the first one to report the carrier frequency of thalassemia and SCA among antenatal women from Telangana population. This study warrants the need for screening antenatal women for these disorders during their early trimester in order to reduce the incidence of affected births.

1. Introduction

Hemoglobinopathies are inherited autosomal recessive conditions affecting the quantity and quality of haemoglobin in red blood cells. Of these, beta thalassemia (β-thalassemia) and Sickle cell anaemia (SCA) are most prevalent requiring lifelong blood transfusions and extensive medical care. These disorders occur in high frequency in the belt extending from the Mediterranean basin through the Middle-East, Indian subcontinent, Burma, Southeast Asia, and Pacific islands but are now common worldwide due to rampant migration.[1,2] They present a significant health problem with approximately 7% of the world population being carriers.

India is considered to be the world capital of thalassemia. Without treatment, their spleen and liver become enlarged and are prone to infections and heart failure which are the leading cause of death among children with thalassemia major. Sickle cell anaemia is the most common blood disorder in India, next to thalassemia that requires lifelong management and contributes to infant and childhood morbidity and mortality. Patients having only SCD are unaware of the disease as they are asymptomatic and

* *Corresponding author*.
E-mail address: sumanjaindr@gmail.com (S. Jain).

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can tolerate Hb levels of 7 or 8g/dl. The key difficulty arises only when the damaged sickle erythrocytes occlude the microcirculation blocking the blood vessels and cause episodes of severe pain. Due to late diagnosis, the treatment of the children is delayed leading to severe pain crisis and other complications resulting in death.

Therefore, numerous screening programs to identify carriers have been proposed as prevention is better than cure. Screening of pregnant women in antenatal clinic is the best suited program in larger towns and cities of India because many pregnant women come for antenatal check-up. All those antenatal women who turn out to be carriers for thalassemia and SCA can be counselled and their partners screened for these disorders followed by prenatal diagnosis (PND) which is acceptable and effective strategy for controlling them in developing countries like India.

Prevention of hemoglobinopathies has been included in government program but not many studies have been conducted and no data has been reported so far on the prevalence of thalassemia and SCA among antenatal women from Telangana population. Hence, the present study was initiated to screen the antenatal women for thalassemia and SCA carrier status during their early trimester (≤20 weeks of pregnancy) to create awareness and know the prevalence of these disorders in the population of Telangana and thereby reduce the incidence of these disorders in the population by preventing the birth of affected children.

2. Materials and Methods

2.1. Subjects

The present study was initially started as a pilot study at Modern Government Maternity Hospital (MGMH), Petlaburz, Hyderabad with approval from the Institutional Ethics Committee (Ref No. IEC/OMC/2021/M.No (01)/Acad-09). The study was later extended to Primary health care centre (PHC), Balanagar and Primary Health Care Centre, Rajapur in Mahbubnagar district (Ref No. IEC/GMCMBNR/AP/08.2021) as the prevalence of thalassemia was found to be high in Mahbubnagar as revealed by our previous study.[3]

A total of 2478 antenatal mothers who are ≤20 weeks of pregnancy are recruited so far in the present study after informing to them the need for screening for thalassemia and SCA. Written consent was obtained from all the subjects participating in the study.

2.2. Collection of blood sample

3-4 ml of venous blood sample was collected from all the antenatal women after explaining to them the importance of the study. Information pertaining to demographic, medical and family history and consanguinity were recorded using a specific proforma. The samples were transported to Thalassemia and Sickle Cell Society (TSCS) for laboratory analysis.

2.3. Laboratory analysis

The blood samples were analysed for Complete Blood Count (CBC) for haematological indices followed by High Performance Liquid Chromatography (HPLC) for determining haemoglobin levels (HbA, HbA2 HbF and HbS) for diagnosing thalassemia (HbA2 > 3.5%) and SCA (30-40%) carrier status.

In case of antenatal women, who were found to be carriers for thalassemia or SCD their partners were counselled and screened. If both the partners were carriers, then they were counselled about the one fourth risk of bearing an affected child at every pregnancy and were advised to go for PND (Chart 1).

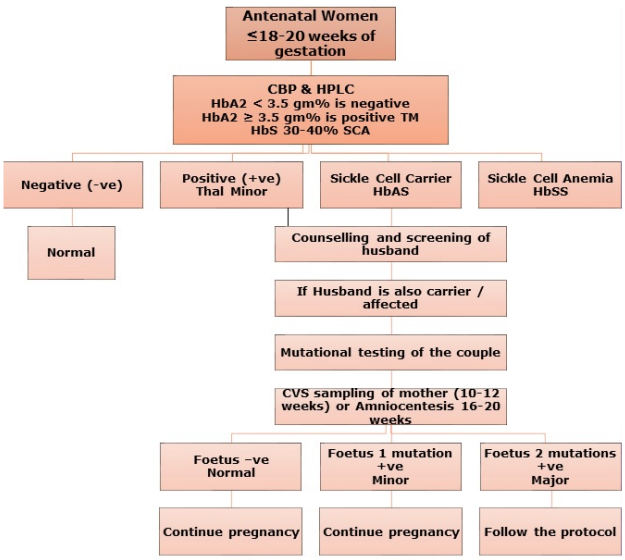

Chart 1: Flow chart for Antenatal screening

2.4. DNA analysis and prenatal diagnosis

If both the partners were found to be carriers, then their DNA was tested for mutation in beta globin gene (*HBB*). After the detection of mutation, chorionic villus sample (at 10-12 weeks of pregnancy) or amniotic fluid (at 16-18 weeks of pregnancy) of the fetus was drawn and analyzed for DNA mutations. If the fetus was found to be homozygous for DNA mutation in *HBB* gene, then the couple was counseled about bearing an affected child and advised to follow the protocol which is legally accepted.

3. Results

A total of 2478 antenatal women (≤20 weeks of pregnancy) are recruited so far in the present study after creating awareness among the antenatal mothers about the prevalence and complications of thalassemia and SCA and

the need for getting screened for these disorders.

Out of 2478 antenatal mothers recruited from MGMH (2000), Rajapur PHC (224) and Balanagar PHC (254) a total carrier frequency of 3.05% (61) and 4.6% (22) was found for different types of hemoglobinopathies from MGMH and PHC's respectively. Table 1 shows the carrier frequency of different types of thalassemia detected in the study. Of the different types of thalassemia, beta thalassemia was the predominant one with a frequency of 2.15% from MGMH and 2.92% from the PHC's. Sickle cell anaemia was also detected with a carrier frequency of 0.55% in MGMH and 1.26% in PHC's of Mahbubnagar. Three couples (4.9%) out of 61 recruited at MGMH were found to be thalassemia carriers. The three couples were counselled about the risk (25%) of having an affected child and were sent for PND on their willingness and written consent. The fetus in all the three cases was found to be normal, and they are continuing with the pregnancy.

Table 1: Carrier frequency of different types of thalassemia among antenatal women

Carriers	**MGMH (N=2000**	**PHC's (N=478)**
Thalassemia	43 (2.15%)	14 (2.92%)
Sickle Cell Anaemia	11 (0.55%)	6 (1.26%)
Delta Beta Thalassemia	1 (0.05%)	1 (0.21%)
HbD Punjab	3 (0.15%)	-
HbE Carriers	1 (0.05%)	1 (0.21%)
Sickle Beta Thalassemia	1 (0.05%)	-
E Thalassemia	1 (0.05%)	
Total	61 (3.05%)	22 (4.6%)

4. Discussion

Hemoglobinopathies impose a significant health problem involving financial, social and emotional constraints. It is estimated that 1.1% of couples are at risk for having children with a haemoglobin disorder, and 2.7 per 1000 conceptions are affected worldwide. The cost of ideal treatment for one thalassemia or SCA child is nearly Rs. 1, 25,000 to 2, 00,000 per annum including the cost of blood transfusions, transfusion pump, leucocyte filters, blood test, hemograms, serum ferritin, liver and thyroid function tests, viral testing, chelation therapy with deferoxamine or deferiprone or deferasirox and hepatitis B and pneumococcal vaccines which is beyond the reach of many parents leading to financial constraints. Furthermore, though prevention of hemoglobinopathies is an established government program, not many studies have been conducted and no data has been reported so far from the population of Telangana. Therefore, an attempt to prevent and control thalassemia and SCA in the population deserves a high priority.

Prenatal diagnosis for beta thalassemia in Chinese resulted in an approximately 70% decrease in the number of newborns affected with thalassemia.[4] Prevention programs for beta thalassemia and sickle cell anaemia in Greece involving antenatal women significantly reduced the number of new affected births.[5] In Taiwan, the national screening program resulted in a marked decline in the number of newborns with thalassemia major.[6] In India, many studies have reported the success of antenatal screening followed by PND. Of the 14,086 women screened in a large maternity hospital in Mumbai, 270 (1.96%) were thalassemia carriers and 22 other hemoglobinopathies.[7] In another study by Colah et al,[8] of 61,935 women screened, 1020 (1.6%) were found to be thalassemia heterozygotes and 213 were heterozygous for other hemoglobinopathies. A study by Bhukhanvala et al,[9] revealed a prevalence of 3.38% beta thalassemia carriers and 1.5% SCA among antenatal women. Baxi et al.,[10] reported 2.78% thalassemia carriers among 1006 women screened at a single centre in Indore, Madhya Pradesh. Shukla et al,[11] reported a thalassemia carrier frequency of 2.95% and 0.05% SCA among 2000 women screened at Department of Pathology in collaboration with the Department of Obstetrics and Gynaecology of a hospital in Delhi from 2010 to 2013.

In the present study a carrier frequency of 2.15% was observed for thalassemia from MGMH, Petlaburz which majorly constituted urban population residing in Hyderabad and Rangareddy districts of Telangana. Unlike MGMH, the PHC's had rural population from Mahbubnagar district comprising of tribal communities and a higher carrier frequency of 2.92% was observed for thalassemia. Prevalence of SCA also was more in the rural population than in the urban as reflected by the frequencies which were 1.26% from PHC's and 0.55% from MGMH. Our findings are consistent with other findings wherein the thalassemia carrier frequency ranged from 2-4% in different populations of India. The frequency of SCA was more in rural population as compared to urban as reported by many studies that SCA is more predominant in tribal groups than in non-tribal ones. A total of 4.9% of the couples screened were found to be carriers and went for PND. This is the first study reporting the carrier frequency of thalassemia and SCA among antenatal women from Telangana population. Keeping in view the high frequency of thalassemia and SCA carriers, this study warrants the need for screening antenatal women at large for these disorders during their early trimester in order to reduce the incidence of affected births and the burden on the families, society and the health care system.

5. Conflict of Interest

The authors declare no conflict of interest.

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Author biography

Padma Gunda, Research Scientist

Saroja Kondaveeti, Medical Officer

Mamata Manne, Research Scientist

Anitha Appam, Assistant Professor

Suman Jain, Chief Medical Research Officer
and Secretary https://orcid.org/0000-0001-6237-1913

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