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FACTORS INFLUENCING BETA-THALASSEMIA AWARENESS IN WESTERN INDIA

Ashwin P Patel[1], Prakash H Parmar[2] , Rupesh B Patel[3], Nikhil M Trivedi[4], Nileshkumar A Bhartiya[5]

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Author's Affiliation:
[1]Consultant Hematologist; [2]Executive director; [3]Consultant Pathologist, Indian Red Cross Society, Ahmedabad; [4]Liaison officer, Hematology Clinic, Hematology Clinic, Ahmedabad; [5]Assistant Camp Manager, Indian Red Cross Society, Ahmedabad

Correspondence:
Patel Ashwin P
rutviap@hotmail.com

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ABSTRACT

Context: Beta-thalassemia is highly prevalent in western India. Our organization runs a screening and prevention program to create awareness and reduce the incidence of homozygous beta-thalassemia cases.

Aims: The study was designed to evaluate factors influencing awareness about Beta-thalassemia.

Methodology: This **c**ross sectional survey was conducted at six colleges, two medical clinics, and a thalassemia transfusion centre. It involved 398 adults (≥18 years).The survey form was objective and written in the local language. Statistical analysis was done using Chi square test in SPSS software version 20.

Results: All the participants knew that beta-thalassemia affected males and females equally. Correct responses to various questions varied from 6.5 % to 45.2 %. The participants with a positive family history or who were already tested for beta-thalassemia had significantly more knowledge ($p < 0.0001$), but even participants with first-degree relatives of Beta-thalassemia major didn't have complete knowledge. Age and gender had no impact while education influenced only certain aspects.

Conclusions: Cascade screening should be used to enhance already increased awareness in people with a positive family history. Aggressive screening of college students will be helpful as they are likely to use it in the near future reducing the incidence of homozygous beta-thalassemia.

Key-words: beta-thalassemia, screening, prenatal diagnosis, hypochromic microcytic anemia

INTRODUCTION

Beta-thalassemia is the commonest genetic disorders[1]. Various strategies are adopted for prevention: population screening[2,3], mass awareness program[4], antenatal screening[5, 6], premarital screening[7, 8], cascade screening[9] and students' screening[10]. Success of any program depends on the culture, education, and circumstances of the target population[8,10,11,12,13,14]. Awareness about thalassemia is one of the many factors that influence thalassemia program. Various factors were identified to affect outcome of thalassemia prevention program. Someone in the family/relatives with thalassemia increases awareness[2]. Premarital screening for preventing marriages between high risk couple may not help as noted in a study from Saudi Arabia[8]. Sometimes patient and their parents hide the information about the disease to avoid social stigma thus preventing awareness, cascade screening of near family members, and acts as a barrier for the program[9]. Awareness program targeting school children may not help as children tend to forget the knowledge acquired by the time they marry [9].

India has a high prevalence of hemoglobinoapthies[3, 15]. There is no national policy about hemoglobinoapthies and hence there are different and

fragmented efforts [3, 6, 16, 17]. There are very few research articles in the Indian literature about factors affecting thalassemia awareness and hence we decided to undertake this study. We wanted to find out factors influencing awareness about beta-thalassemia so that our screening program can be modified accordingly.

SUBJECTS AND METHODS:

This cross-sectional survey was carried out in six colleges, two outdoor medical clinics, and at a thalassemia transfusion centre. These places were selected to include all sections of the society including relatives of the patients with beta-thalassemia. Only adults (≥18 years) were included in the survey. The form was kept simple, objective, and in the local language so that it could be filled out quickly (Figure 1). The ethical committee approved the study. Informed written consent was taken from each participant. The participants were counseled once they completed the survey to increase their awareness. The survey was designed to gauge the knowledge about the preventive aspect of beta-thalassemia and not the diagnostic /treatment part of the disease. Subjects with positive family history or who had undergone thalassemia screening in the past were expected to have more awareness and so the subjects were divided in to four groups for the analysis purpose as follows:

Group 1= all participants
Group 2= participants with first degree relatives suffering from β-thalassemia major
Group 3= Participants who already had β-thalassemia test
Group 4= Participants without positive family history and/or did not have previous β-thalassemia test

Chi-square was used for statistical analysis (SPSS software version 20) and a p-value of < 0.05 was considered as significant.

RESULTS

Table 1 gives the demographic details of the participants (n=398). The average age was 38.1 years (range: 18-80 years) and 30.2 years (range 18-62 years) for male and female participants respectively. The education level was comparable between females and males. Table 2 mentions the correct response percentages for each survey question. The question "Is thalassemia a genetically transmitted disease?" received maximum positive responses (45.2 %) while very few participants knew about the need to test cousins of a thalassemia patient (6.5 %). The survey form had separate questions for husband, wife, uncle, aunt, maternal cousins, paternal cousins, brother, sister, father, and mother but there were equal positive responses for husband/wife, father/mother, uncle/aunt, maternal cousins/paternal cousins, and brother/sister and hence for the analyses purpose the categories were clubbed as spouse, parents, uncle/aunt, cousins, and siblings respectively.

Table 1: Demographic details of the study participants (n=398)

Demographic Profile	Participants
Sex (n=398)	
Male	245 (61.6)
Female	153 (38.4)
Mean Age in years (range)	35.2 (18-80)
Education (n=355)#	
Undergraduate:	170 (47.9)
Graduates	184 (51.8)
No education	1 (0.3)
Family history (n=326)*	
Thalassemia minor	10 (3.1)
Thalassemia major	72 (18.1)
First degree relatives	59 (14.8)
Already tested for β-thalassemia (n=372)$	107 (28.8)

43 subjects did not reveal their educational status
* Includes subjects with only family history of thalassemia minor (excludes subjects with family history of thalassemia major)
$ 26 subjects did not answer this question

Table 2: Positive responses (percentage) in various groups

Question	Group 1 (n=398)	Group 2 (n=59)	Group 3 (n=107)	Group 4 (n=267)
Knew/heard about β-thalassemia	59	96.6	96.3	42.3
People knew that β-thalassemia was genetically transmitted	45.2	86.4	77.6	30.3
People knew that β-thalassemia could be prevented by prenatal diagnosis	38.9	79.7	67.3	26.6
Spouse of a person with β-thalassemia should be tested for β-thalassemia	31.4	83.1	61.7	16.1
Parents of a person with β-thalassemia should be tested for β-thalassemia	25.4	39	42.1	18.4
Siblings of a person with β-thalassemia should be tested for β-thalassemia	23.9	76.3	43	7.9
A person with low hemoglobin should be tested for β-thalassemia	21.4	44.1	40.2	12.4
Uncle/aunt of a person with β-thalassemia should be tested for β-thalassemia	15.6	20.3	15.9	2.6
Cousins of a person with β-thalassemia should be tested for β-thalassemia	6.5	71.3	41.1	1.9

Group 1= all participants; Group 2= participants with first degree relatives suffering from β-thalassemia major; Group 3= Participants who already had β-thalassemia test; Group 4= Participants without positive family history and/or did not have previous β-thalassemia test

Table 3: Comparison of positive responses according to gender and educational level

Responses	Gender			Education		
	Male (n=245)	Female (n=153)	P value	Less than graduation (n=170)	Graduation or more (n=184)	P value
Who should be tested for β-thalassemia?						
A person with low Hb %	48 (19.6)	37 (24.2)	0.33	27 (15.9)	50 (27.2)	0.03
Parents of a person suffering from β-thalassemia	55 (22.4)	40 (26.1)	0.47	38 (22.4)	57 (31)	0.08
Siblings of a person suffering from β-thalassemia	57 (23.3)	38 (24.8)	0.81	48 (28.2)	42 (22.8)	0.06
Husband/wife of a person suffering from β-thalassemia	68 (27.8)	57 (37.3)	0.06	60 (35.3)	60 (32.6)	0.02
Uncle/aunt of a person suffering from β-thalassemia	15 (6.1)	9 (5.9)	0.90	12 (7.1)	11 (6)	0.92
Cousins of a person suffering from β-thalassemia	36 (14.7)	26 (17)	0.64	44 (25.9)	15 (8.2)	<0.01
Heard about thalassemia	145 (59.2)	90 (58.8)	0.97	95 (55.9)	117 (63.6)	0.20
knew about genetic transmission	102 (41.6)	78 (51)	0.09	79 (46.5)	90 (48.9)	0.02
knew about pre-natal diagnosis	87 (35.5)	68 (44.4)	0.09	77 (45.3)	69 (37.5)	<0.01

There was no statistical difference when the responses to various questions were compared between male and female participants. Education had varying levels of influence; people with lower education (up to high school) had more knowledge about the need for spouse testing and prenatal diagnosis compared to the group with higher education (graduation or post graduation) while people with higher education had more knowledge about genetic transmission and the need to check for beta-thalassemia when hemoglobin was low (Table 3).

People with positive family histories of beta-thalassemia had significantly higher knowledge ($p < 0.001$ for all the questions). Table 2 gives the breakup of the responses given by the people who had a first-degree relative suffering from Beta-thalassemia major (group 2). People who were already tested for beta-thalassemia also had higher knowledge (Table 2, group 3) ($p < 0.001$ for all the questions). People with no family history or no previous beta-thalassemia testing had very little knowledge (Table 2, group 4). Positive family history was the most likely factor that motivated a person to undergo beta-thalassemia testing ($P < 0.001$). Young undergraduate females with no family history of beta-thalassemia were tested more frequently.

DISCUSSION

Beta-thalassemia awareness programs are shown to increase the knowledge amongst the at-risk population [4, 18]. This is the first study, to the best of our knowledge, which has analyzed the factors influencing the knowledge of Beta-thalassemia. The study has brought out many important observations that will guide future awareness programs. People with positive family history and people who had undergone beta-thalassemia screening (CBC and HPLC) had significantly more knowledge about Beta-thalassemia ($P < 0.0001$). The people who were tested for beta-thalassemia were always counseled during the screening program, thus explaining their higher knowledge. The positive impact of counseling on the knowledge level is also previously reported [2, 5]. The knowledge increased even when there was a positive family history in only distant relatives. However, it was surprising to know that the knowledge and awareness were far from the expectation when there was a positive family history of thalassemia major in first-degree relatives (Table 2). High percentage of people knew that beta-thalassemia is transmitted genetically (86.4 %), and that prevention is possible with prenatal diagnosis (79.7%). There were also good awareness about who should be tested in the family; 76.3 % of the people knew that siblings should be tested, 83.1% knew that spouses should be tested, and 71.3 % knew that cousins should be tested. In contrast, only 39% of people surveyed knew that parents should be tested, 20.3% knew that uncle/aunt should be tested, and 44.1% knew that low hemoglobin levels could be due to Beta-thalassemia. A previous study also has shown that thalassemia status is not always shared with the family members [19], although this is changing [20]. Our study was not designed to analyze the knowledge sharing amongst the family members and relatives but this could be one of the reasons for lower awareness in this high-risk group. Another reason could be denial towards the hereditary nature of the disorder among family members as reported previously [21]. It would be interesting to follow this group and see how much knowledge is retained later on, as knowledge tends to wear off with time [2]. A previous study from India has shown that the knowledge was not retained even in subjects with beta-thalassemia minor [10].Regular counseling at frequent intervals is needed to help retain the knowledge or screening needs to be done during times when the knowledge is more likely to be used in the near future, for example, near the marriage period.

All the participants knew that Beta-thalassemia affected males and females equally; there were equal positive responses for husband and wife, for uncle

and aunt, and for paternal cousins and maternal cousins. Interestingly, there was higher knowledge in the group with less education (middle school/high school education) about the need for spouse testing and prenatal diagnosis. We initially thought this to be due to the targeting of college students in our awareness program. But when analyzed further, it was found that majority in this group were not current students (68.8 %). This group is probably more receptive to the teaching of awareness programs compared to the population with higher education. A person with a positive family history of beta-thalassemia major was more likely to be tested for thalassemia, as seen in our study, as he /she has seen the suffering involved with the disease. In contrast to the normal belief and previous reports [22], there was no impact of participants' education level on the screening test being performed which could be due to the impact of our aggressive awareness program.

Although our study shows that people with positive family histories had more knowledge about beta-thalassemia and that they were more likely to undergo thalassemia screening, the knowledge was not sufficient (Table 2). A previous study has also shown that even after extensive counseling, only 43% persuaded their near relatives for thalassemia testing and therefore we can conclude that all the near relatives need separate counseling [2] .Cascade screening (screening of extended family members of a person with beta-thalassemia major/minor) is a more cost effective form of screening as a higher percentage of the at-risk population can be identified, saving health resources in the process [9, 16].

In spite of aggressive thalassemia awareness program in our region, the knowledge of beta-thalassemia was not very impressive in the group that has had no previous screening for beta-thalassemia or a positive family history; 30.3% of people surveyed knew that thalassemia was genetically transmitted while only 1.9% knew about the need to test cousins of a thalassemia patient (Table 2). It is probably worth targeting a high-risk population (relatives of a patient with thalassemia minor/major, pregnant anemic females, and college students) rather than targeting the whole population during awareness/screening program. Antenatal screening has been reported from different parts of the world, including India, to be highly effective [5, 6, 16, 23].

The males and females in our study had equal knowledge about thalassemia, which is in concurrence with a previous report [22]. Young undergraduate females with no family history of thalassemia opted thalassemia screening more frequently. This could be due to targeting of college students in our screening program and more receptiveness of females to awareness campaign.

We have not studied whether higher knowledge of thalassemia leads to reduction in marriages of couples with Beta-thalassemia minor or increase in prenatal diagnosis and termination of pregnancy if a fetus is found to be homozygous. However, previous reports have documented reduction in marriages when both partners had beta-thalassemia minor and increase in termination of pregnancies when fetus was found to have beta-thalassemia major [5, 6, 7, 16]. Thus, it can be presumed that the incidence of beta-thalassemia is expected to fall with increased awareness. The study was not designed to find out the impact of different programs (antenatal program, screening of college students, population screening, etc.) or the different components of a program (lectures, one-to-one counseling, media campaigns, distribution of leaflets, etc.) on the knowledge; however, this type of data may help in designing cost effective programs.

This study will help in changing strategies in the future. People knew about the genetic transmission but were not sure about who needs screening for beta-thalassemia in the family when a member was found to have Beta-thalassemia. This is crucial to find out prevalent cases of beta-thalassemia minor. Placing more importance on cascade screening and counseling about autosomal recessive transmission will be helpful.

In clinical practice, more importance is given to the screening of childbearing relatives of the affected population, i.e. siblings, cousins, and spouse, and less importance is given to the screening of parents and uncle/aunt. This is reflected in the lower knowledge about the need to test parents and uncle/aunt. Cascade screening and counseling about autosomal recessive transmission will be helpful.

There is less awareness that beta-thalassemia is one of the common causes of anemia. This is one of the reasons of missing beta-thalassemia minor status of a pregnant female leading to the birth of a beta-thalassemia major child. Expansion of antenatal screening, prenatal diagnosis, and placing more stress about this aspect in the awareness program will be helpful.

A positive family history and testing of thalassemia had a substantial impact on the awareness. Awareness in remaining population was very low in spite of aggressive awareness and screening programs in our region for the last ten years. It will be worth targeting at-risk groups (antenatal screening, cascade screening, screening of college students) to be more cost-effective.

CONCLUSIONS

There was significantly less awareness in spite of existence of thalassemia awareness program for last 10 years. Positive family history and previous enrollment in thalassemia screening had significant impact on the knowledge indicating that personal exposure about the disease severity and counseling during thalassemia testing were very helpful. The awareness was not as expected even when there was family history of thalassemia (majority had thalassemia major) in first degree relatives indicating that more needs to be done. Cascade screening, repeated counseling, and emphasis on the weak areas of counseling (like stress on mode of genetic transmission, suspecting thalassemia when hemoglobin is low) can help.

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