

Parental Awareness and Cascade Screening of Thalassaemia in Sri Lanka

Abstract

A strategy to prevent new births with thalassaemia major in countries where abortion is illegal is to screen the at risk population for the carrier state and advocating "safe marriages". One of the highest risk groups to have thalassaemia gene are the relatives of individuals with a child with 'Thalassaemia major'. 'Cascade screening' of such relatives is an effective way to initiate and precede a much wider population screen. We wanted to identify the level of awareness of disease transmission and the extent to which cascade screening was already happening in Sri Lanka. 200 parents of children with transfusion dependent thalassaemia attending the Thalassaemia units at Ragama and Kurunegala were interviewed. Seventy three percent fathers and 91% mothers knew their carrier state. 59(64.1%) fathers and 98(88.1%) mothers knew their carrier state has caused the illness to the child. Mother had 382; fathers had 291 siblings and those siblings had 714 and 610 children respectively. 40 (10.8%) of maternal siblings and 15(5%) of the paternal siblings had been screened for thalassaemia. Only 48(6.72%) of maternal side children and 23 (3.77%) of paternal side children has been screened. Wider awareness and a screening programme targeting those high risk populations must be launched without delay because prevention is better both for the patient aspect as well as the cost aspect to the government. More studies probably qualitative needed to evaluate the individual factors for above results.

Keywords: Safe marriages; Thalassaemia; Cascade screening; Implementation; Population

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Nilanga Nishad AA^{1*} and Premawardhena AP²

¹Biyagama Ministry of health, Sri Lanka

²Department of Clinical Medicine, University of Kelaniya, Sri Lanka

***Corresponding author:** Nilanga Nishad AA, Medical officer of health, Biyagama, Ministry of health, No; 122/A/3, Maharanugegoda, Ragama, Sri Lanka, Tel: 094718331470; Email: aanilanga@gmail.com

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Introduction

In countries such as Sri Lanka; where abortion is illegal, strategies to prevent new births with thalassaemia major is limited to screening the at risk population for carrier status and advocating 'safe marriages'. The 'safe marriage' strategy ensures that neither or only one of the partners is a beta-thalassaemia carrier at marriage or when the decision to bear a child is made. Cascade screening of relatives of thalassaemia major patients is considered as a way to initiate and precede a much wider population screening of thalassaemia carrier status [1]. Cascade screening involves the screening of all immediate relatives of both (parent) arms of a thalassaemia major child. Yet, some scientists believe that we can never reduce the thalassaemia burden unless a policy change favouring induced abortions occurs. We tried to identify the level of awareness of disease transmission among parents of thalassaemic children and to estimate the extent to which cascade screening had already penetrated Sri Lanka before the universal screening program began.

Methods

We recruited 200 parents of transfusion dependent thalassaemia patients from two main and two peripheral thalassaemia centres in Sri Lanka using stratified random sampling and interviewed both mother and father. A self administered questionnaire was developed in Sinhala language to collect data on parents' knowledge on their carrier status and to assess the extent to

which carrier screening had taken place in their families. Review of medical records was conducted in addition to the interviews. Data were analyzed for percentages and comparisons were done to determine which arm of the family was better screened [2].

Results

There were 100 parent couples representing 100 transfusion dependent thalassaemia children. The mean age of thalassaemic children was 13 years (SD= 7.3). In the assessment of knowledge regarding the thalassaemia, 91% mothers and 73% of fathers were aware of their carrier status. Eighty eight percent of mothers and 64% of fathers knew that their carrier state had predisposed their children to the illness. Table 1 shows the extent to which siblings of the parents and the nuclear family members of those siblings had been screened for thalassaemia. As in Table 1, significantly higher number of siblings from the mothers' side (10.8%) had been screened for thalassaemia than from the fathers' side (5%). More children of siblings from the mothers' arm (6.7%) were screened than those of the fathers' (3.8%) as well. Interestingly, more spouses of siblings from the fathers' arm (6%) had been screened than the mothers' arm (3.6%). All these differences were statistically significant at 1% level.

Discussion

In Sri Lanka, there seem to be deficiencies at present in the screening of relatives of patients with thalassaemia. Unfortunately,

locally or internationally there are no other published studies to compare these findings with. A probable reason for poor level of penetration in cascade screening could be the social stigma, limiting the parents from divulging their child's thalassaemic condition even to their closest relatives. According to Sangani et

al. [3], 20% of families with thalassaemia children had expressed having had unfavourable reactions from their relatives. Non availability and non-affordability of screening facilities was also described as a limiting factor identified in India. These same reasons may have affected the Sri Lankan populations as well.

Table 1: Status of thalassaemia screening among relatives of parents with thalassaemia.

Parent	Mother's Arm	Father's Arm	Total	Significance
Number of siblings	382	297	679	P <0.01
Number of siblings screened	41	15	56	
Percentage	10.80%	5%	8%	
Number of spouses of the siblings	334	248	582	P <0.01
Number of spouses screened	12	15	27	
Percentage	3.60%	6%	4.60%	
Number of children of the siblings	714	610	1324	P <0.01
Number of children screened	48	23	71	
Percentage	6.70%	3.80%	5.30%	

Conclusion

However, wider awareness and screening programs targeting those high risk populations have become the biggest hope in reducing new births with beta and E beta thalassaemia, where abortion is not only considered illegal, but also a sin. This letter also highlights the deficiencies of cascade screening as a program to reduce the disease burden, and may justify implementation of programs such as essential pre marital thalassaemia screening.

References

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