

Safe Marriage for Thalassaemia Prevention; the Need for Public Health Education

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Rec date: Oct 13, 2015, Acc date: Dec 10, 2015, Pub date: Dec 18, 2015

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Abstract

Thalassaemia instigates an enormous health problem all over the world, due to the palliative nature of life long treatment and the cost involved. The available preventive strategies are not simple and not universally acceptable. The concept of "safe marriage" proposed for thalassaemia prevention in Sri Lanka recommends to ensure that one partner in a couple be a non-carrier. Thereby the option of screening at least one of the partners becomes the minimal essential requirement for a safe marriage. The process of screening, counselling and decision on selecting partners is voluntary and health education programmes only promote this. However to establish such a practice in society, the wide spread practice of screening and counselling should be established. Stringent monitoring to evaluate and take remedial action in the programme would also be essential.

Keywords: Thalassaemia; Marriage; Health education

Introduction

Thalassaemia (ICD-10 code: D56) is a chronic haemolytic disorder caused by a genetic defect in the production of either beta (β) or alpha (α) globin chains of the haemoglobin molecule. In thalassaemia, patients have defects in α or β globin chain, leading to production of abnormal red blood cells. A wide spectrum of genetic defects resulting in either qualitative or quantitative deficiencies of either one of the globin chains, gives rise to diverse outcomes and clinical pictures. Some of the affected babies die prematurely and the other extreme do not manifest any clinical illness [1,2].

In Sri Lanka, as in other South Asian countries, beta thalassaemia predominates. Even though a higher prevalence is reported in three provinces where malaria has been rampant in the past, carriers who amount to 2.2% of the population are distributed all over the country [3]. At least 80-100 thalassaemia babies are born to the birth cohort of 300 000 babies every year giving an annual incidence of 0.03%. With the survival rate of about 25 years the country has a prevalence of 2000-3000 thalassaemia patient. This number is on the increase due to lack of a prevention program [4].

Management of beta thalassaemia involves regular and lifelong blood transfusions and chelation therapy to remove iron, which accumulates due to regular blood transfusions. The human body is not equipped with a mechanism of removing excess iron. Therefore accumulated iron has to be removed by regular chelation therapy. Blood transfusion combined with iron chelation is very effective in prolonging life; the patient almost achieves normal life expectation. However, the longevity achieved by such expensive state sponsored free treatment is confounded by the escalation of the cost of care adding burden to the already strained health budget of the country. The estimated cost of 5% of the health budget is an underestimate of the gravity of the problem, as it does account for the personal expenses of individuals. Patients and families face enormous challenges and pains

except for free transfusion services facilitated by generous blood donors in the country [5].

Thalassaemia prevention in Sri Lanka

The mainstay of thalassaemia prevention in many countries is based on pre-natal diagnosis and abortion [6]. However, in Sri Lanka and many other countries, abortion is prohibited by law. Lack of facilities for chorionic villi biopsies adds on to the problem. Efforts at legalizing abortion as an antidote to thalassaemia could not see the light of the day owing to social, religious and cultural beliefs. Therefore, the concept of safe marriage as a preventive measure has been proposed. This approach is heavily dependent on population screening for thalassaemia carriers. Thalassaemia carriers are asymptomatic. Unless they are screened, they have a risk of culminating in an 'at risk' marriage by chance. Therefore, teenage and adolescent screening is the only simple and beneficial solution directed at the criteria recommended by the Wilson and Jungner principles of population screening [7].

Thalassaemia carriers are asymptomatic. However as they have a significant risk of ending up in an 'at risk' marriage, population screening would be beneficial. As the screening test is simple, cost effective and the available solution is also practical, the population screening has become very rational and adheres to all criteria recommended by the Wilson and Jungner principles of population screening [7].

A marriage is defined as safe when one partner is a non-carrier for thalassaemia. This concept stands true for any recessively inherited disorder. In thalassaemia the idea becomes realistic as the carrier state could be recognized by a simple blood test, a full blood count (FBC) by an automated haematological analyser. The national thalassaemia prevention program of Sri Lanka promotes voluntary screening of teenagers and adolescents according to screening protocol (Figure 1).



Figure 1: Thalassaemia horoscope; an aid to decide whether a proposed couple is safe or at the risk of having offspring with thalassaemia [9,10]. Box number 2 and 4 is highlighted to indicate that those who select a known thalassaemia carrier as their partner in marriage should check their blood by HPLC to confirm that they are non thalassaemia traits and counselling them regarding selecting a partner for marriage [8,9].

The counselling process is facilitated by the concept of horoscope reading using a pictorial depiction of a matching couple and at risk couples simulating a horoscope (Figure 2).



Figure 2: National Thalassaemia Screening Protocol of Sri Lanka [9,10].

According to the national thalassaemia screening protocol, a mean corpuscular volume (MCV) less than 80 fl and a mean corpuscular haemoglobin (MCH) value less than 27 pg provides reasonable assurance to exclude possible thalassaemia carrier state. They would be given a green card and be recommended no restriction in selecting a partner for marriage as there is negligible risk for offspring being affected with thalassaemia [8,9]. The word negligible is used in place of no risk as there is a chance of missing an e beta thalassaemia carrier and a rarer chance of two such carriers meeting in marriage [8,9]. Those who have microcytosis are treated with iron by the general practitioner over a period of three months and the blood test is repeated. If the microcytosis has disappeared the cause of the microcytosis would be recognized as iron deficiency and they also would be given a green card with the same assurance. Those with persisting microcytosis are likely to be thalassaemia traits. Therefore they would be given a pink card and counselled not to couple with another red cardholder or someone who has not been screened. Thalassaemia carriers need much more careful counselling and support to address their stresses and possible stigma in society [8,9].

Population screening for thalassaemia involves the entire society and this will create a significant workload load for primary care doctors. As the screening test detects thalassaemia trait as well as iron deficiency, a large number of populations will be positive for the initial test. This is an advantage for societies with high prevalence of iron

deficiency, ranging 5-20% in some regions. However, first contact doctors would be burdened with these patients who need counselling and education.

Public education and provision of facilities for screening and counselling is essential. The general public is aware of the disease and over 95% of the public has recommended screening at least one of the partners in a previous study. However, establishing the practice of screening [8] and decision making regarding selecting partners has not been successful, even after 10 years of accepting the policy [9].

The thalassaemia prevention program with voluntary screening and counselling is unlikely to be successful unless there is a proper monitoring system. Screening coverage, incidence of high-risk marriages and incidence of at risk pregnancies and incidence of thalassaemia at national and regional level would be valuable indicators to monitor the success of such prevention programmes [9].

Conclusions

'Safe marriage' concept; the thalassaemia preventive strategy in Sri Lanka could be adopted in any nation and for any autosomal recessive disease. In those countries with an established practice of antenatal diagnosis, safe marriage would be another practical option to offer for a person who has not decided on a marriage partner. The pictographic depiction of the carrier status and at risk marriage will be a useful aid for counselling carriers of autosomal diseases.

References

1. Olivieri NF (1999) The beta - Thalassaemia. *The New England Journal of Medicine* 341: 99-108.
2. Rund D, Rachmilewitz E (2005) Beta-thalassemia. *N Engl J Med* 353: 1135-1146.
3. de Silva S, Fisher CA, Premawardana A, Lamabadusuriya SP, Peto TE, et al. (2000) Thalassaemia in Sri Lanka: implications for the future health burden of Asian populations. Sri Lanka Thalassaemia study group. *Lancet* 355: 786-791.
4. http://www.thalassaemia.org.cy/wp-content/uploads/pdf/communication-awareness/TIF%20Magazine_Issue%2060.pdf.
5. Mudiyanse RM (2006) Thalassaemia treatment and prevention in Uva Province, Sri Lanka: a public opinion survey. *Hemoglobin* 30: 275-289.
6. Angastiniotis MA, Hadjiminias MG (1981) Prevention of thalassaemia in Cyprus. *Lancet* 1: 369-371.
7. Wilson JMG, Jungner G (1968) Principles and practice of screening for disease. Public Health Paper, Number 34. Geneva: WHO.
8. Mudiyanse RM. Safe marriage for prevention of thalassaemia: Sri Lankan experience, appraisal of the concepts and challenges of implementation. *J Pharm Biomed Sci* 2015;05(10): 791-796.
9. Mudiyanse RM, Senanayaka MP, Rathnayaka RMS (2015) Safe Marriage for Thalassaemia Prevention. KAP survey in Sri Lanka. *Translational Biomedicine* 3: 26.