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Awareness Of Beta -Thalassemia In Tamil Nadu, India: Llustrated By 2 Case Scenarios: Urgent Need For Increasing Awareness

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Abstract

Background: Beta thalassemia is an autosomal recessive inherited genetic disorder which is emerging as an important disease with a huge economic burden on India. Health policy makers need to recognize this and plan strategies for effective awareness and prevention programs throughout the country and not just the tribal areas.
Case report: We present two families with more than one child with beta thalassemia major and discuss the factors regarding the recurrence. In the first family, both children were siblings while in the second family they were cousins.
Results: Both the families were not aware of the existence of thalassemia before the diagnosis in their children in the first family. The delay in diagnosis was due to the late presentation of the disease. In the second family there was a lack of awareness of the genetic nature of the disease which prevented the family from seeking preventive measures.
Conclusion: Awareness about beta thalassemia and the screening facilities available should be popularized by policy makers so that people are empowered to use them without any stigmatization or discrimination.

Keywords: ***Beta Thalassemia, Awareness, Screening, Prenatal Diagnosis.***

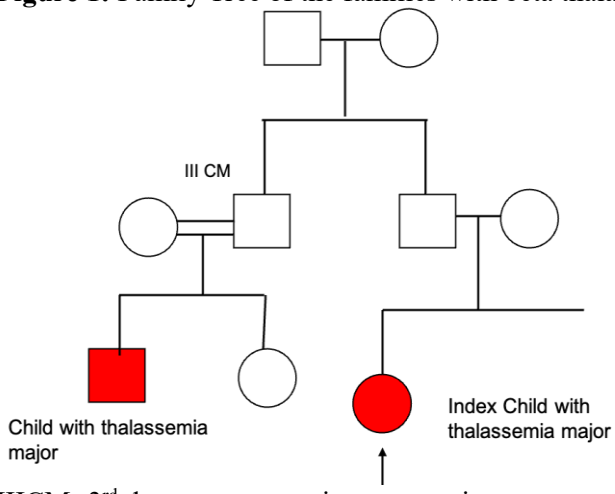
1. INTRODUCTION

Case report:
We present the case report of two families who have siblings or cousins affected with thalassemia major, who had attended the hematology department at Voluntary Health Services, Chennai.

Family1:
This was a III-degree consanguineous couple (marriage between first cousins) whose first child was diagnosed with thalassemia major at 5 years of age. The family hailed from Muslim community and were from Chennai, the capital of Tamil Nadu. At 5 years, the elder child became very lethargic following a bout of respiratory infection and febrile illness. During evaluation, her hemoglobin was 3.5g/dl, RBC count -2.56 million/microlitre, Mean Corpuscular Volume (MCV) -68 fl and Mean Corpuscular Hemoglobin(MCH)- 23.8 pg. Her Hemoglobin variants estimation by HPLC showed HbF (Fetal hemoglobin) to be 93.5%, HbA -5 and HbA2 1.5 confirming the diagnosis of beta thalassemia major. Subsequently the child received packed cell transfusions every 3 weeks to maintain her hemoglobin around 9g/dl. The child underwent a successful bone marrow transplant and is doing well now. She is 9 years old. The younger sibling who was 3 years at the time the elder sibling was diagnosed, was asymptomatic. She was screened by complete blood count and HPLC and was also diagnosed to have beta thalassemia major. Her hemoglobin level at 3 years was 11g/dl. HPLC showed HbF to be 92.1 %. She continued to be asymptomatic until 5 years when she developed anemia following respiratory infection. She needs packed cell transfusions every 3 weeks and is currently awaiting bone marrow transplantation. Both children underwent molecular testing of the HBB gene and were found to be homozygous for the common mutation IVS1-5 (G>C) also called as HBB:c.92+5 G>C and the parents were heterozygous carriers. The parents were educated but had never heard about thalassemia before their first child was diagnosed and had not undergone premarital or antenatal screening. Unfortunately as the disease presented late in the elder child, prevention of disease was not feasible in the second child.

Family 2:
Family 2 was a nonconsanguineous Hindu couple from Dharmapuri- Harur and they have settled in Chennai. Their first child was diagnosed to have beta thalassemia major. She was 6 months at the time of diagnosis. Parents noticed that the child had become pale, lethargic and had abdominal distention. On evaluation her complete blood counts revealed hemoglobin of 4.7 g/dl, RBC count was 1.94 million/microlitre, MCV – 68.6 fl, and MCH -24.2 pg. Hemoglobin variants estimation by HPLC revealed HbA-7.6%, HbA2 -2.1% and significantly elevated HbF of 87.9%, confirming the diagnosis of beta thalassemia major. Molecular testing of HBB gene revealed homozygous IVS1-5 (G>C) mutation also called as HBB: c.92+5 G>C and the parents were heterozygous carriers. The parents were counselled about the genetic nature of the disease, the need for regular transfusions, the availability of bone marrow transplantation and the feasibility of prenatal diagnosis. The family also informed that the husband's brother's child was also reported to have severe anemia in infancy and had been receiving blood transfusions regularly. The husband's brother has married his own first cousin. However further details were unavailable on that visit. Subsequently, the family was able to retrieve and share the child's reports. On perusing the records, the brother's child was also found to have beta thalassemia major. The parents were very upset and wished that they could have been informed about the possibility of screening for this disorder earlier. The husband's brother's family were also counselled about the genetic nature of the disease. Consanguinity increases the risk of sharing of common gene pool of not only beta thalassemia but also other recessively inherited disorders . Both the families were explained that the carrier state of beta thalassemia in India is 3-4% and therefore even in non-consanguineous couples can have children with beta thalassemia major.

Figure 1: Family Tree of the families with beta thalassemia major



IIICM- 3rd degree consanguineous marriage

2. DISCUSSION

Beta thalassemia is one of the most common monogenic disorders worldwide and in India too. With an estimated burden of 1,00,000 patients with thalassemia major and around 10,000 children being born annually, it is very difficult to optimally manage children in a resource constrained developing nation like India[1]. The ideal way would be prevention of beta thalassemia by creating awareness, identifying carriers and motivating couples to undergo prenatal diagnosis. Beta thalassemia is an autosomal recessive disorder, hence if both the partners are carriers there is a 25% risk of having a child with Beta thalassemia major. Another important factor is the presence of consanguineous marriages[2,3]. Consanguineous marriages are deep rooted in India, especially in South India. The main reasons being financial and emotional security for the women and ensuring that the assets remain within the larger family. With increasing educational status and better living conditions, the rate of consanguinity is declining [4,5,6]. Nevertheless, the percentage of consanguineous marriages is the highest in Tamil Nadu and is estimated to be around 28% while the national average is around 11% according to the latest National Family Health Survey -5 by the Government of India[4].

Micro mapping in various areas have shown that certain communities and tribal populations in Northern parts of India have a carrier frequency of 5-17%. However not much information is available about South India especially Tamil Nadu. With the decline of infective and nutrition related disorders, inherited disorders are becoming more significant and beta thalassemia being foremost. It has been estimated by the National Health Mission that 10,000-15,000 babies are born with thalassemia major every year in India. This disorder can be currently treated by Bone marrow transplantation, but the major limiting factor is the availability of HLA matched donor and the exorbitant cost. Hence the mainstay of treatment for the majority of children remains regular blood transfusions and iron chelators to prevent iron overload [1,2]. The lifelong treatment of these children in an efficient manner places a huge economic burden on the country's economy. In several countries like Israel and Cyprus, there are effective screening programs for prevention of beta thalassemia and it was found that prevention was greatly cost effective in comparison to treatment[1]. The main cornerstone of prevention is creating awareness. Without a proper awareness any screening program is likely to create more anxiety, stigmatization and refusal to accept the program. In our case history, family 2 clearly shows the lack of awareness of the disorder even when a close relative has been affected. In the first family routine screening by complete blood counts and Hemoglobin HPLC would have helped in identifying carriers. Screening programs can help these families to a great extent. But screening should be voluntary and there should be good pretest and posttest counselling session and guidance towards prenatal diagnosis [7].To start with, screening of pregnant women registered with health care system and then following up with screening the spouses of those women who were found to be carriers would help in identifying the families who would need prenatal diagnosis. As Tamil Nadu has a good healthcare network this can definitely be carried out with success. But the crux would remain to create adequate awareness through multimedia and social media so that people take up screening voluntarily.

3. CONCLUSION

Universal screening for beta thalassemia is of paramount importance in India. Screening for beta thalassemia does not involve technically challenging molecular genetics. It can be done by tests that are available easily including CBC and HPLC. This should be offered to all people at least in antenatal population to begin with, and in addition a meticulous follow up of results with a posttest counseling will eventually help in identifying and preventing Beta thalassemia in the society. As heterozygotes are silent carriers, voluntary screening, counseling and avoidance of marriage between carriers will go a long way in reducing the incidence of beta thalassemia in the country.

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Conflict of interest: None

Data availability: The raw data supporting the conclusions of this article will be made available by the authors without undue reservation Written informed Consent was obtained from the participants As this was a case report without revealing patient identity, ethical approval was not obtained

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