What and why you need to know about beta thalassemia: A genetic disorder

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ABSTRACT

Thalassemia is a genetic disorder. Now a day's beta thalassemia is becoming quite prevalent all over the world. The prevalence rate is varied in the different state of India. Beta (β)-thalassemia is of three types. The beta thalassemia major, minor and inter-media. The beta Thalassemia major patients have to be dependent on regular blood transfusion to sustain life. Only BMT (Bone Marrow Transplantation) is the cure for these patients which are costly in the developing country. The condition significantly impacts patient quality of life and imposes substantial healthcare costs, exceeding 1 lakh INR annually for treatment and management.

The role of genetic factors, such as polymorphisms in MTHFR, prothrombin and Factor V Leiden, has been studied extensively, although results remain inconclusive. Societal factors, including the rise in inter-caste marriages without genetic screening, contribute to the increasing prevalence of thalassemia. Preventive measures, such as pre-marriage, antenatal and neonatal screening, are essential for early detection and management. Universal implementation of such screenings, as practiced in countries like Greece and Italy, has significantly reduced disease incidence.

India has initiated measures, such as free carrier screening, blood transfusions, and HPLC testing, particularly in states like Uttar Pradesh. However, integrating affordable and accessible Point-of-Care (POC) diagnostic tests is crucial for timely intervention. Comprehensive efforts combining governmental policies, genetic counseling, and public awareness are vital to combating this genetic

disorder.
Keywords: Beta thalassemia; Genetic disorder; Bone marrow transplantation

INTRODUCTION

The approximate cost of treatment and management of thalassemia patients cost more than 1 lakh a year. The beta thalassemia major patients frequently suffer from many vascular problems. Thrombophilia is a blood disorder that comprises imbalances in the blood coagulating factor due to ecological and hereditary components. Previous evidence shows that thrombosis is the commonest risk in beta-thalassemia patients [1,2]. Several studies have examined that MTHFR C677T, prothrombin G20210A (PT G20210A) and Factor V Leiden G1691A (FVL G1691A) polymorphism play a crucial role in the development of β-thalassemia major, yet the result was questionable and uncertain. Therefore, in this study, we executed the correlation between these gene polymorphisms with β-thalassemia major patients [3].

Regarding the treatment and management of the thalassemia patient we the state of Uttar Pradesh is blessed to have a great support from the blood cell division of National Health Mission, UP, India.

DESCRIPTION

Still the new mutations are emerging due to the mixing up of the gene pool. We need to be more focus in the diagnosis of genetic disorder. Earlier the main concern was in the particular community like Bohras, Muslims etc. One major reason to pop up the cases of thalassemia major is due to inter-caste marriage without seeing the genetic status of the individual. Earlier matching the gotra was the main issue before marriage [4]. Now days in India we say first match HBA2 status to check thalassemia status.

The government of the country should make various screening and diagnosis mandatory for the eradication of thalassemia. They are indicated as under mentioned [5]. The country Greece has followed these rules now Greece is a thalassemia free country: Pre-marriage screening, antenatal screening, preconception screening, neonatal screening, preoperative/pre-anesthesia screening and genetic counseling.

Pre-marriage screening: Pre-marriage screening should be implemented to detect β thalassemia carriers and hemoglobinopathies such as sickle cell trait. In developing country, it is not frequently acceptable because of social stigma and reasons in the general public.

The pre-marriage screening should be possible in universities and colleges, schools or public places such as theater, shopping mall etc. Earlier it was performed in where the predominance of thalassemia is high. But nowadays due to the mixing up of the gene pool this screening is recommended in all colleges where students are of marriageable age. Our lab has performed this screening in the Tharu tribal area of the KheriLakhimpur, Uttar Pradesh, India. We have collected more than 700 samples from the college and carrier screening of sickle cell disease has been performed. It is a belt of HBS [6].

Antenatal screening: In antenatal screening irrespective of gestational age of all pregnant women should be screened for carrier status of thalassemia and hemoglobinopathies. The spouse of the affected female should be test for mutation such as α-, β thalassemia and hemoglobinopathies (HbS trait, Hb E trait, Hb D trait etc.) [7].

The prenatal diagnosis have to be encouraged if the foetus is in danger for the having thalassemia mutations e.g. α-, β thalassemia and hemoglobinopathies (HbS trait, Hb E trait, Hb D trait etc.). If the couple found positive for thalassemia during antenatal screening they can decide for prenatal diagnosis and subsequent pregnancy [8-11].

Preconception screening: While troublesome circumstances are in the developing country like India, this ought to be done but mostly females frequently do not enroll in antenatal centres before 12 weeks of gestation. A similar methodology with respect to antenatal screening ought to be followed. The preconception screening is important for all couples coming to IVF in infertility clinics. If the female is a thalassemia carrier then her husband or sperm donor should be screened and vice versa [8-11].

Neonatal screening: Infant screening is mainly suggested in haemoglobinopathies such as sickle cell diseases, prevalent in tribal and urban populations. If possible, neonatal screening has to be implemented universally where infants are at high-risk for homozygous β-thalassemia and all instances of HbS-β thalassemia. This methodology will miss a couple of instances of sickle β-thalassemia when the mother is a β-thalassemia carrier and the father is a carrier

of HbS. All babies with significant hemoglobinopathy should be re-tested using molecular technology to confirm the diagnosis within three months after birth [8-11].

Preoperative/pre-anesthesia screening: Preoperative/pre-anesthesia screening of patients is preventive measure where the prevalence of HbS is high. As the presence of sickle hemoglobin might interfere during preoperative and postoperative procedures [8-11]

Genetic counseling: Genetic counseling should be given by an expert to affected thalassemia families. The advice can likewise be given by a trained genetic counselor, a hematologist or a pediatrician [8-11].

Point of Care (POC) testing for thalassemia

As there is a saying that prevention is better than cure. The thalassemia and hemoglobinopathies are genetic disorder and due to the migration of population in the different regions and endogamy the new combinations of thalassemia hemoglobinopaty are arising fast. For the eradication and further treatment and management of thalassemia disease an effective diagnostic test at the Point of Care (POC) is the need of hour. As thalassemia has been spread worldwide an early diagnosis, economical test, awareness programmes and prenatal screening will be a milestone for the eradication of this genetic disorder and to reduce burden of the health sector of a country subsequently the economics. The initial hematological, biochemical screening to the advance molecular testing less than one roof will not only help to diagnose the thalassemia patients but shall be also helpful in the treatment and management of the disease. The objective of POC for testing thalassemia will be achieved if these quick testing methods like NESTROFT. This has to be in the approachable distance to the patients.

CONCLUSION

The government should include the thalassemia screening as a mandatory tool for screening. The best example Italy and Cyprus there with the initiation of prenatal diagnosis and screening of the carrier now this is a thalassemia free country. Likewise in India, the Uttar Pradesh state government has initiated the task by giving free of cost of screening of thalassemia carrier, blood transfusion and iron chelators to several medical colleges. The Indian government has provided the HPLC machines to provide the screening of the carrier of thalassemia hemoglobinopathies. Basically the eradication, treatment and management of thalassemia are joint efforts of government, stake holder, policy makers, pediatrician, pathologist, transfusion medicine and geneticist.

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