Prevention of Thalassemia Major: Need of the time

All parents wish to have healthy children. There are many genetic diseases with serious consequences , for example, thalassemia, hemophilia, intellectual disability and others. Children with these disorders can be born in any family. Prevention of genetic disorders is an important antenatel care.

Prenatal investigation

Is the baby in my womb alright? It is impossible to answer this question. But thanks to our progress in science and technology, we can detect many genetic diseases in the unborn child. Thalassemia can be diagnosed in the baby in womb (fetus). Termination of pregnancy if the fetus is found to be affected with Thalassemia major is the way to prevent the birth of a child with thalassemia major in the family.

Problem of beta thalassemia in India

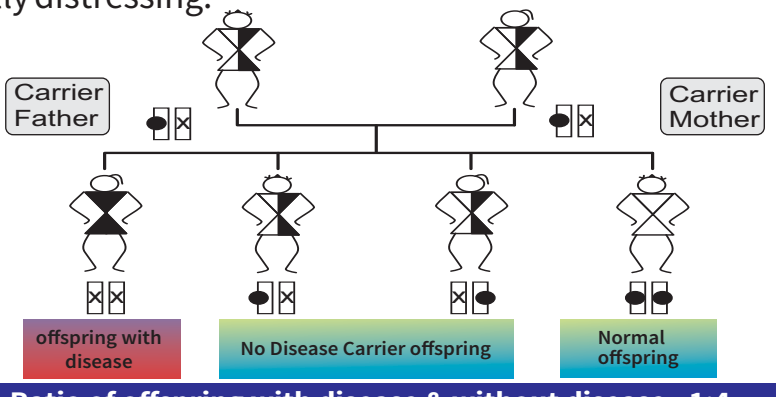
Beta thalassemia is a common genetic disease in our country. Three out of 100 people are carriers of the beta thalassemia variant. In other words anybody can be a carrier of beta thalassemia. A carrier is a person who themselves do not have any disease but their children are at risk of having thalassemia major which is a serious illness. About one in thousand couples is at a risk of giving birth to a child with thalassemia major.

What is thalassemia major?

Thalassemia major is serious disease presenting with severe anemia [lack of blood] between the ages of 6 months and 2 years after birth and if not treated timely, they succumb to the disease usually by one to five years. A child with Thalassemia major can be born only if both of the couple are carriers of beta Thalassemia or other hemoglobinopathy. During each pregnancy the possibility that the child will have thalassemia major is 25% [i. e. 1 in 4]. The possibility that the child born to a couple who are carriers of beta thalassemia will be free from thalassemia major is 75% [i. e. 3 in 4].

What is the treatment of thalassemia major?

This life threatening disease can only be cured by bone marrow transplantation (BMT). This is possible only if matched donor, preferably sibling is available. Another approach to treatment is regular blood transfusion (usually every 2 to 3 weeks) along with chelation therapy. However this treatment not only puts a financial burden on the affected families but also is emotionally distressing.



Ratio of offspring with disease & without disease - 1:4

Can we prevent birth of a child with thalassemia major?

Yes, prenatal testing can be done between 12-20 weeks of gestation to detect Thalassemia major in the fetus. If the fetus is found to be affected, the family may consider the option of termination of pregnancy to avoid the birth of a baby with thalassemia major.

Can there be birth of a child with thalassemia major in my family?

A child with Thalassemia can be born to any couple. If both the parents are carriers of beta Thalassemia and / or other hemoglobinopathy like Hb E, S, C, etc. then the risk of having a child with thalassemia major is 25% in every pregnancy. Anybody can be a carrier and carrier detection can be done by a simple blood test. If parents are found to be carriers then the family may opt for prenatal testing.

What is the test to detect carrier of beta Thalassemia? Where is the testing available?

Carrier detection can be done at the department of Medical Genetics, SGPGIMS, Lucknow by a blood test known as HPLC of hemoglobin. It provides levels of Hb A2, Hb F and any other variant hemoglobin if present. In addition red cell indices including mean corpuscular volume (MCV) and mean corpuscular hemoglobin (MCH) should be done. Our department is committed to create awareness about population based screening for beta thalassemia for control of thalassemia. Carrier detection is best done before pregnancy or within first 2 months of pregnancy. This allows for timely intervention and assurance of a thalassemia free child. DNA based prenatal diagnosis for thalassemia major is a modern and accepted technique and, in Uttar Pradesh this facility is available only at SGPGIMS, Lucknow.

Prevention is better than treatment

Thalassemia major is a difficult and serious illness to treat but its prevention is easy. Common people and obstetricians must know about the need of population based screening and prenatal diagnosis to fully utilize the potential of this facility. Awareness about this service can prevent many families from dreadful and unfortunate consequences of thalassemia major.

Timely detection of families at risk of birth of other genetic disorders by drawing a pedigree is essential to provide prevention of genetic disorders with serious consequences.

Points to note

Carriers of beta thalassemia do not have the disease; nor does being a carrier lead to any other health problems. *Most families who have a child with thalassemia major usually do not have family history of thalassemia major. *Thalassemia carrier status is not a stigma and a person cannot be blamed for being a carrier of beta thalassemia. *Knowledge regarding thalassemia is meant to make you vigilant to prevent the birth of a child with thalassemia major. * Screening of children should be avoided. Pre-marriage screening need not be encouraged and is not necessary.

Birth of a healthy infant makes the family exultant

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If available and affordable, pre-pregnancy screening for Spinal Muscular Atrophy (SMA) and Fragile X syndrome also may be offered to women planning pregnancy