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Genetic carrier testing is the key to reducing occurrence of Thalassemia

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Even as medical science strives to find a cure for thalassemia, making genetic carrier testing more prevalent among would-be parents can drastically reduce the occurrence of this inherited disease in the population

On the eve of World Thalassemia Day, the scientific team at the Bengaluru-based genomic diagnostics and research firm, MedGenome Labs, stressed that ALL couples getting married should undergo carrier testing and genetic counselling routinely, and this is even more critical in the case of Thalassemia.

Even as medical science strives to find a cure for thalassemia, making genetic carrier testing more prevalent among would-be parents can drastically reduce the occurrence of this inherited disease in the population.

Thalassemia is an inherited (genetic) blood disorder in which the body makes a reduced quantity of haemoglobin. It is one of the leading causes of severe haemolytic anaemia amongst infants which is associated with excessive number of red blood cells being destroyed by the body, requiring lifelong multiple blood transfusions to stay alive.

Thalassemia affects both males and females uniformly and is prevalent across all states in India. Marriages between close members of a family or between members of close-knit local communities is responsible for at least 3-17% of thalassemia occurrences in India.

Dr. Sheetal Sharda, Clinical Geneticist with MedGenome Labs said, “Would-be parents, who are carriers of the disease, are at a greater risk of having a child with Thalassemia Major, especially if they are married to another person who is also a carrier. A couple with Thalassemia trait, have 1 in 4 chances of having a child with Thalassemia Major even if they don’t show any symptoms themselves or have no family history.”

In the current Indian context, advanced genetic testing for carrier screening of parents, combined with genetic counseling, is the best way to reduce the occurrence of thalassemia in India. Genetic analysis can help the couple take informed reproductive decisions for their future married life. There is an urgent need to launch a national campaign to popularize genetic testing of ALL individuals and not only those with a family history, to reduce the healthcare burden on our society due to untreatable genetic disorders", she added.

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There is no cure for thalassemia currently. Treatments include blood transfusions, iron chelation therapy (removal of excess iron from the body) and bone marrow transplant. Blood transfusions at intervals of 2-3 weeks are prescribed to patients affected with Thalassemia Major. This can be emotionally and financially draining for the family. The best solution is to prevent it from happening and all it takes is a screening test of the couple when they are planning for a family. The screening test is fairly simple and is now accessible for people across the country.