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Reduce passing on the faulty gene to the next generation

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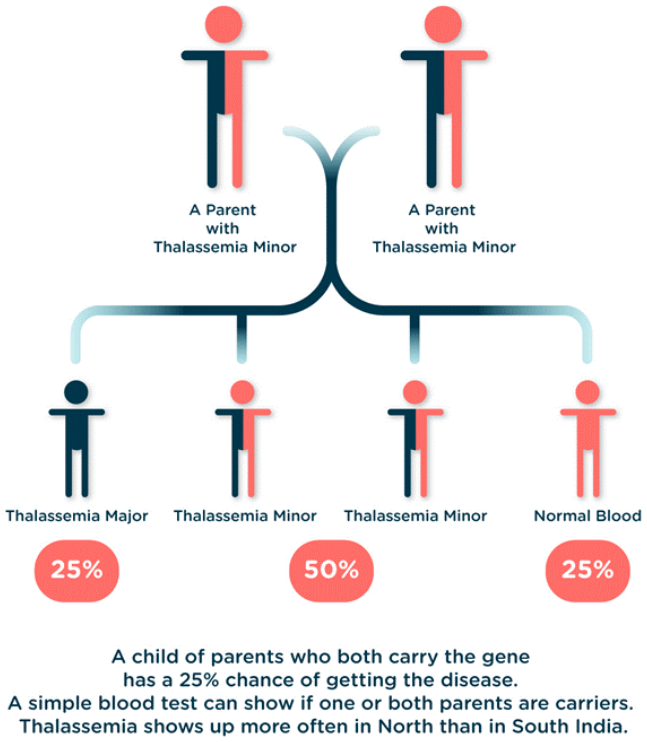
Of the thousands of IVF procedures that are performed each year, 70% are performed for couples who have difficulty conceiving. The remaining 30% of the IVF procedures are associated with Pre-implantation genetic diagnosis (PGD).



India reports an average of 10,000* children born with Thalassemia major every year. The disease which is inherited is passed on to 25 % of the offspring when both the parents are carriers of the mutated gene. This development of the abnormality in the embryo happens at the conception stage, hence, the parents are not aware whether the child is at risk as it poses no health issues to them. Awareness plays a major role in Thalassemia which is currently lacking in most couples. Child with Thalassemia is called Thalassemia major and requires blood transfusion at regular intervals and the only approach to cure this hemoglobin disorder is bone marrow transplantation with suitable donors.

Today, with the advancements in science and medical care, couples who are at risk of carrying inherited diseases can take to Pre-Implantation Genetic Diagnosis (PGD) Testing to help reduce the chances of passing on the faulty gene to their child.

PGD testing happens after the IVF procedures and is an effective method to detect and rule out specific diseases and abnormalities in the embryo. An abnormal embryo can lead to miscarriage, fetal death later in the pregnancy or sometimes result in stillbirth. Post the fusion of gametes in IVF, the health of the embryos are checked through PGD. Pre-implantation genetic diagnosis is a method of studying the chromosomal balance and identifying the presence of a genetic defect in an embryo before it is implanted into the womb. This helps in improving the chances of a healthy pregnancy and baby.

Human leukocyte antigen (HLA) typing (a system used for matching bone marrow donors) along with PGD helps in conceiving a child who is disease free and can donate cord blood or haematopoietic stem cells for transplantation to save an affected sibling. Haematopoietic stem cell transplantation (HSCT) from related matched donors improves overall survival compared to unrelated or non-matched donors. Since HSCT from related matched-donors is unavailable for 70% of patients, IVF for PGD-HLA is a relevant clinical option. This can help couples give birth to a savior sibling for saving an older child affected by the disease.

What is thalassemia and how is it caused?

Thalassemia is a form of anaemia and results in chronic tiredness, bone problems, spleen problems, heart disease and more. A genetic disorder which an estimated 8,000 – 10,000 babies are born every year with in India.

Because, it is an inherited disease, when both parents are carriers of the gene, but not suffering from the disorder (people like this are known as having thalassemia minor), it can be passed on to the children. Affected children are referred to as being thalassemia major.

Treating Thalassemia

For less severe cases, nutritional supplementation and regular blood transfusions are some of the ways of treating and managing the condition. There are other therapies too, but, especially in younger children, bone marrow transplantation from a matching donor helps overcome the disease in 80-90% of the cases.

The problem with this is finding a matching donor, one who's HLA (Human Leucocyte Antigen) is identical with the patient's. Internationally, thanks to 22.5 million donors, the possibility of finding a match for an Indian patient is 16%. But in India, because there is just over 30,000 donors the chance is a measly 0.008%!

Management of infertility requires a thorough work-up, including pre-pregnancy counseling of the couple. Fertility assessment of patients with thalassaemia should also include evaluation of the partner according to standard criteria. The fertility options are dependent on two factors (a) partners' carrier status and (b) site of damage to the H-P-G axis.

If both partners are homozygous for ?-thalassaemia, use of donor gametes, preferably donor sperm is the ideal option, as sperm can be more easily available from sperm banks, whereas the use of donor eggs is technically more complicated with an unpredictable success rate . If the partner is heterozygous, then **P**re-implantation **G**enetic **D**iagnosis (**PGD**) is another option, where diagnosis can be made prior to conception. This method may be more acceptable to certain communities with religious beliefs against termination of affected pregnancy. Lastly, in patients with severe organ damage or where both partners have HbTh, another option may be adoption, where there is good family support.

At Nova IVI Fertility, we implement the latest in medical technology to help our patients fulfill their dreams of having a healthy child and prevent foreseeable diseases in their children.

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