Maya, who hails from a small town in Karnataka was married, as per tradition, to her first cousin Arjun (her father's sister's son) and experienced the unexpected. Her first born daughter was born with a genetic disorder. Tragically, the baby died within four days of her birth. Totally ignorant about what engendered this, Maya and Arjun had another baby who unfortunately met with the same fate. If this couple would have known the cause of death of their first child, they could have accordingly taken preventive steps for their second child.

In another case, a non-consanguineous couple lost one child on the 52nd day of life. The baby was suspected to be affected with maple syrup urine disorder. Maple syrup urine disease (MSUD) is an inherited genetic disorder in which the body is unable to process certain amino acids properly, resulting in a severe neurological disorder which could be fatal in the time of crisis. The child could not be investigated further due to early demise.

If the parents in such cases get screened for genetic disorders, the chances of the child inheriting it can be reduced greatly. But the Indian society, in general, is still unaware about genetic disorders and their consequences. Majority of these disorders are untreatable and create a lot of emotional and financial burden on the family, and society. Prevention is the only way forward to stop these disorders from passing down in the generations. Carrier screening is one such test which can identify disease-causing variations in people and make them aware of their risk of passing on the abnormal gene to their progeny.

What is a carrier?

A carrier is a person who is mostly asymptomatic i.e. without any physical manifestation of the disorder, but he/she has the ability to pass on his/her altered or defective gene to the offspring. Prenatal testing, in case of positive family history, has proved to be beneficial for preventing the birth of an affected child. But even in cases of a negative family history, the couple can still seek genetic counselling and get themselves tested and prevent the disorder in the family. Prevention is better than cure for many genetic disorders.

Need for screening

Each individual is a carrier for an average of 2.8 known severe recessive mutations. The most commonly inherited genetic disorders in India include hemoglobinopathies, congenital adrenal hyperplasia, glucose-6-phosphate dehydrogenase deficiency, neuromuscular disorders, inborn errors of metabolism, and cystic fibrosis.

Carriers are usually healthy or unaffected but they have a risk of passing on a genetic condition to their children. Traditionally, carrier screening has been offered to patients based on their ethnic background or family history; but more than 80 per cent of babies born with inherited genetic diseases have no known family history.

Every individual inherits two copies of each gene, one from each parent, in case of autosomal genes. Carriers have one copy of the defective gene and the other is normal. Two copies of the mutated gene must be inherited for an autosomal recessive disorder. If both the parents carry the same defective gene, then there is a 25 per cent chance that the child will be born with a genetic disorder.

Carrier screening is of great importance as it determines the risk of conceiving a child with an inherited disorder. It also helps in early diagnosis, timely intervention (prevention, management, treatment), markedly reduces disease severity, improves quality of life, substantially benefits psychosocially, may prevent death by intervention and most importantly gives an opportunity to the parents to make an informed decision.

In the second case where the baby was diagnosed with MSUD, the couple was advised carrier screening for the presence of the genetic defect. On