analysing the parents' DNA sample, each of them was found to have a significant mutation in the DBT gene, which is a known to cause MSUD. In the next pregnancy, prenatal diagnosis in the form of chorionic villus sampling at 12-13 weeks was offered. The foetus was found to be unaffected, as both the mutations found in the parents were absent in the foetus. The couple continued the pregnancy and were able to prevent a second baby with the same disorder.

Thus, testing for the mutations, found in the parents by screening, is helpful in preventing such disorders in the future generations.

Who needs to be screened?

Any person, irrespective of family history or ethnicity, can be screened for carrier status of genetic disorders. But there may be some who are at high-risk:

People with a positive family history of genetic disorders should get screened. Since they have seen someone close suffer from such a disorder, they know the consequences and often approach the clinician to know the risk of the ailment in their next child.

Marriage within relatives or close family, better known as consanguineous marriages, are common in some communities and states. They become a major factor for children being born with genetic disorders as they have a greater chance of sharing the same genes as their family members.

A community with a high risk of certain disorders should consider such tests to make informed choices. Certain communities do not marry outside their closed groups and they are at high risk of genetic disorders due to inbreeding.

How to go about it?

Couples who are planning to get married (premarital) or planning a baby (pre-conceptional) or have just conceived (post-conceptional) should ideally should opt for this test. Opting for a session of genetic counselling will be very helpful in understanding the family history, ethnicity and consanguinity and accordingly test for certain disorders. It is suggested that the couple take this test together which requires only a simple blood sample. In case the result for a genetic disorder is positive, the doctor or the counsellor will explain in detail the reproductive options for the couple.

Carrier screening for a couple dramatically reduces their chances of having a child with a genetic disorder. It provides the couple an opportunity for reproductive choices, including pre-implantation genetic diagnosis, prenatal diagnosis and preparedness for the newborn. With advancement in science, you can use next generation sequencing to get screened for all recessive diseases with high accuracy at an affordable cost, which is a big boon.

So, instead of horoscope matching, why not opt for genetic matching before tying the knot?

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