What is trisomy?

EXTRA CHROMOSOME A type of polysomy, trisomy can cause a variety of genetic disorders. Dr Priya Kadam elaborates on some of them

Most people have 46 chromosomes, 23 inherited from each parent. However, in certain cases, an individual may have 47 chromosomes instead of 46. Having this extra chromosome may lead to physical abnormalities and problems related to intellectual and developmental functions. This is known as trisomy, which means three bodies.

Common conditions

Trisomy is a genetic disorder where in a person has three copies of a chromosome instead of two. Trisomy can occur in any chromosome and often leads to miscarriage. Some of the common conditions are:

- **Trisomy 21:** It is a genetic condition where a person has three copies of chromosome 21. It is also known as Down syndrome and affects approximately one in 800 babies. Individuals with Down syndrome have mild to moderate intellectual and developmental disabilities, heart defects along with other medical conditions.
- **Trisomy 18:** It is a genetic condition where a person has three copies of chromosome 18. This is also known as Edwards's syndrome and it affects approximately one in 3,000 babies. Infants with Edward's syndrome have severe intellectual and developmental disabilities along with other birth defects that make them less likely to live beyond a year.
- **Trisomy 13:** It is a genetic condition where a person has three copies of chromosome 13. This is also known as Patau syndrome and it affects approximately one in 5,000 babies. Infants with Patau syndrome have severe intellectual and developmental disabilities, serious heart problems and multiple physical problems that make them unlikely to live beyond a few weeks.

Trisomies of other chromosomes such as 16, 8, 9 and 22 are incompatible with life and often lead to spontaneous miscarriage during first or second trimester.

Apart from these, trisomy can also occur in chromosomes X and Y leading to XXX (triple X syndrome), XXY (Klinefelter syndrome) and XYY.

Most of these conditions occur spontaneously in any pregnancy. However, the risk of carrying a baby with these abnormalities increases with maternal age.

Screening

Non-invasive prenatal test (NIPT) is one of the best ways for an expectant mother to check the baby's health without an invasive procedure. A simple blood sample from the pregnant woman's arm is used to examine foetal DNA to rule out any possibility of defects. Other tests which can also determine these defects are chorionic villus sampling (CVS) and amniocentesis, which are invasive.

While there is no cure for trisomy yet, parents of affected children can get guidance from support groups.

(The author is programme director, MedGenome NIPT, Bengaluru)