

Some women should test for Down syndrome

Down syndrome, or Mongolism, is associated with certain facial characteristics, IQ scores ranging from 25 to 75, and abnormalities of heart, bowel and other organs. Down syndrome is due to chromosomal problem. A healthy individual has 23 pairs of or a total of 46 chromosomes. Yet, people with Down syndrome have an extra chromosome in the 21st pair, which comes from mother in over 90% of cases. Translocation or the transfer of genetic material from one chromosome to another accounts for 3% of all cases of Down syndrome.

Women of all age may give birth to a baby with Down syndrome, although the chance increases with age. The older the women, the greater risk of having a baby with Down syndrome.

In the past, pregnant women can undergo an invasive test like chorionic villus sampling or amniocentesis for the detection of Down syndrome before 20 weeks of gestation. Although the accuracy of both tests is near 100%, they are associated with 0.5-1% risk of miscarriage.

Recently, some non-invasive tests are available, including the measurement of fetal nuchal translucency, maternal human chorionic gonadotrophin (hCG) and pregnancy associated plasma protein-A (PAPP-A). Pregnant women can undergo these tests between 11 to 14 weeks' gestation. If thick fetal nuchal translucency, low PAPP-A and high hCG levels are found, the risk of having a baby with Down syndrome will be high and an invasive testing will be indicated.

The following groups of women require prenatal diagnosis and counseling service: (1) age 35 or above, (2) gave birth to a baby with congenital disease, (3) family history of Down syndrome, and (4) any abnormal findings on prenatal ultrasound examinations.