Rapid advancement in the genetic discoveries has increased the knowledge about the role of genes in health, from conception until death. Due to complexity of genetic disorders, affected babies and children cause multisystemic involvement to the family and the individual. Additionally the burden of genetic disorders falls on the health services of every country (Gogate, 2006). It is now possible for people to take an advantage of genetic testing which can identify at risk families and individual through various carrier testing programs. As most of the genetic disorders do not have pre or post treatment, early diagnosis helps in prevention of a serious outcome (Purandarey, 2009).

Human genetic material can be studied in various body tissues like bone marrow, blood, amniotic fluid, chorionic villi and product of conception. This is constitutional and remains same in all the tissues throughout life, except in neoplastic condition (Abramsky, 1994). Genetics has two components one of which studies heredity and variation and the other studies the medical implication and disease with altered genes. The importance of this is in clinical, public health and research (Gardner, 2004).

Prenatal diagnosis deals with management and correction of a defect when possible. (Thasnevis, 1996) The decision of a selective termination, when not treatable and post birth management, if the couple decides to deal with the handicapped child.

Various invasive and non-invasive techniques are available for such diagnosis. Invasive tests are amniocentesis, chorionic villi and cord blood sampling while non invasive tests are ultrasound and maternal serum screening (Phadke, 2007). This study includes the evaluation of chromosomal abnormality from amniocentesis of high risk pregnancies. If the chance of developing genetic disorder increases then such pregnancy referred as high risk pregnancy (Krishna, 1993).
The high risk factors associated with pregnancy are,

**Advanced maternal age:** Advanced maternal age is defined as an increased age at which women give birth to their child. The risk for chromosomal aneuploidy increases with advanced maternal age. The risk associated with advanced maternal age is not only restricted to Down’s syndrome. Screening of women with advanced maternal age also shows aneuploidy for other chromosomes like 13, 18, sex chromosomes or presence of other marker chromosomes (Broke, 1992).

**Increased NT (Nuchal Translucency):** Nuchal translucency (NT) is the redundant skin at the nape of the neck with considerable subcutaneous fat. This observation has been used to increase the detection rate of Down’s syndrome in the first trimester. NT of 3mm or more in the first trimester is considered significant for fetal aneuploidy. In the second trimester NT of 6mm is consider significant (Wieacker, 2010).

**Maternal serum screening:** This is the marker test done it from the blood sample of a risk mother. Markers are the chemicals secreted by the placenta and mainly fetal liver (Crandall, 1991). The test is dependent on the combinations of age, PAPP-A (Pregnancy Associate Plasma Protein type –A), AFP-Alpha Feto Protein, human chorionic gonadotrophin hormones and unconjugated estriol (Broke, 1992).

**Abnormal fetal ultrasound findings:** Many structural abnormalities in the fetus are associated with recognizable chromosomal syndrome (Nyberg, 1990). Known ultrasound marker and associated chromosomal abnormality form an important indication for prenatal diagnosis (Sadler, 2006).

**Genetic disorder in a child, carrier parents:** Fetal chromosomal abnormalities are known to occur due to maternal or paternal non disjunction errors. Couples with a family history or child with a genetic disorder are advised to undergo prenatal diagnosis to assess the carrier status of the couple and to confirm the cytogenetical abnormality present. (Margaret, 1991).
**Bad obstetric history (BOH):** BOH is the case history of couple, who have repeated abortions. There is an increase risk of abnormal child in couple with the history of repeated abortion (Wieacker, 2010).

Steps involved in study are procedure of fetal tissue sampling, culturing of tissue, harvesting, staining-banding and analysis of chromosomal spread.

**Our Hereditary material**

Each cell of our body consists of compact DNA material, which lies as chromosome. When they arrange serially, the complete set is called karyotype. Human have total 23 pairs of chromosomes (Broke, 1992). Chromosomes are of two types i.e., Autosomes and Sex chromosome. Out of total 23 pairs, 22 pairs are autosomal chromosomes which deal with hereditary information, while one pair of sex chromosome which deals with sex chromosomal abnormality (Nussbaum, 2001).

![Normal male karyotype](image1) ![Normal female karyotype](image2) (Purandarey, 2000)

Any alterations in the structure or number of chromosomes refer to chromosomal abnormalities (Dewald, 1983). The abnormalities associated with high risk pregnancies are numerical or structural and are Down’s syndrome, Edward’s syndrome, Patau’s syndrome, Neural tube defects NTD (Milunsky, 1992), Klinefelter syndrome, Turner syndrome, and structural abnormality (Goldstein, 1998 and Crissman, 2011). Prenatal diagnosis also plays an importance role in case of ambiguous genitalia for fetus (Adam, 2012).