Abstract

Rapid advancements in genetics have broadened the knowledge about the role of genes in human 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 the country. It is now possible for people to take advantage of genetic testing which can identify at risk families and individuals 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.

If the chance of developing genetic disorder increases then such pregnancies are referred as high risk pregnancies. 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. This study includes the evaluation of chromosomal abnormality from amniocentesis of high risk pregnancies.

In this study total 1177 cases were enrolled with the high risk factors. Indications were Maternal Serum Screening (MSS)(31.10%), positive maternal serum screening 366 (31.10%), 29 (2.46%) cases with NT, 162 (13.76%) cases with abnormal ultrasound accept NT, 20 (1.69%) cases with BOH, 136 (11.55%) cases with previous child affected and 14 (1.19%) cases of carrier parents. Maternal age was the most common and highest risk factor for which patient referred for amniocentesis followed by positive maternal serum screening and abnormal ultrasound findings.

Among 1177 cases, there was no growth in 12 (1.01%) cases. In remaining 1165 cases, 1081(92.70%) cases are observed with normal karyotype. Chromosomal abnormalities observed in total 85(7.29%) cases, which is much higher then the other studies done before in prenatal diagnosis. If we compare the abnormalities observed, with the world wide abortion risk during amniocentesis, the data highlights the importance of prenatal diagnosis.

The conclusion is made that maternal age is common and highest indication for amniocentesis followed by positive maternal serum screening and abnormal ultrasound markers. Incidence of Down’s syndrome and other chromosomal abnormalities increases with advanced maternal age. Maternal serum screening is usually done for suspected abnormalities like Tri-21, 18 and NTD.
In study the result shows that apart from suspected results unexpected result like structural abnormalities and Trisomy of sex chromosome may also come as a surprise. In study it was observed that the incidence of unexpected results in positive MSS is higher than that of suspected chromosomal abnormalities. In abnormal ultrasound marker, it was observed that USG abnormal markers like absent nasal bone, hypoplastic nasal bone were highly associated with trisomy 21. Increase NT thickness is also associated with trisomy 21. 75% cases observed with Downs’s syndrome were with increased NT. In BOH no numerical abnormalities were observed. The consequences of structural abnormalities observed depends on the rearrangement occurred. In previous affected child the parental karyotype was conducted to rule out the carrier status of parents. For carrier parents, if the fetus carries the de-novo chromosomal patterns then the risk for birth defect or mental retardation or both increases depending on the rearranged chromosomal pattern. i.e De-novo translocation or gain or loss of chromosomal segments.

The incidence of genetic disorders is very low as compared to the other medical conditions, though they are individually rare, collectively more. As there is no definite treatment of genetic disorders, the affected individuals have very poor quality life and there is little awareness about the importance of genetic disorders in healthcare. There are lots of myths, misconceptions, about this in general population. However, they should receive accurate information by appropriate genetic counseling. Identifying the chromosomal abnormality by prenatal diagnosis gives an option to the couple to plan families.