ABSTRACT

The study of genetic basis of pediatric disorders has become one of the most active areas in human genetics. Genetic disorders are enormous, millions of people are suffering from inherited diseases. A very large population of India with high birth rate and consanguineous marriage favored in many communities a high prevalence of genetic disorders. The present study has been made to analyse the genetic basis of a few pediatric disorders like Down syndrome and sex chromosomal aneuploidy in Mysore population. Though many investigators claimed that advanced maternal age is the well established risk factors for causing nondisjunction of the above aneuploidy, there was no systematic study to elucidate the other etiological and demographic factors in India. Therefore, the present investigations were undertaken. Study on the prevalence of pediatric disorders in Mysore revealed 2.06 to 4.45% of abnormal births, of which 0.9 to 2.2% are with genetic disorders. The common genetic disorders among them are Down syndrome, Turner and Klinefelter syndrome. A total of 190 prospective clinically diagnosised Down syndrome and sex chromosomal aneuploidy were used for the present study from three major Hospitals of Mysore and rehabilitation homes. A total of 200 healthy families were also randomly selected from different ethnic backgrounds as controls from different localities of Mysore city, South India. Genetic register was established for 190 patients and 200 controls.

The findings are as follows: Of the 190 patients screened for chromosomal anomalies about 150, 16 and 9 of the patients were shown Trisomy 21, X0, and XXY syndromes respectively, and the remaining mentally challenged patients did
not show any chromosomal anomalies. Of all the Down syndrome patients, 97.6% were with Trisomy 21, 1.6% cases with trisomy mosaic and 0.8% with translocation trisomy. The mothers of control, Down syndrome and sex chromosomal aneuploidy families produced the children in their young age than the advanced age. Advanced age of father and grandmother are the risk factors to cause nondisjunction of chromosome 21, and for nondisjunction of chromosome X grandmother age is the risk factor. Marriages between uncle-niece as well as second-cousins are the possible risk factors for causing trisomy 21. The analysis revealed that the region-wise distribution, educational status, habits of the parents and mother not undergone prenatal diagnosis are some of the other possible risk factors to cause Down syndrome and sex chromosomal aneuploidy.

Counseling will help to have better knowledge about preparation for pregnancy, pregnancy maintenance, prenatal diagnosis and maintenance of genetic disorders like the commonly occurring Down syndrome and sex chromosomal aneuploidy. The best way to reduce the frequencies of any particular genetic abnormality in the population is to reduce the rate of reproduction by those individual capable of having affected offspring.

Thus, to prevent births of unwanted children with anomalies comprehensive maternity care services must be available to all pregnant women regardless of socio-economic status. Therefore, the new born screening and counseling for inheritable diseases in India should be established as a preventive public health programme on a priority basis as immunization program.