Chapter 7

Future Prospects

Various familial aggregation studies and linkage analysis are being embarked on to determine the inheritance pattern of a disease in different genetic disorders in different populations. The results of these studies along with the expected results many a times reveal unexpected associations. Before the advent of chromosomal mapping techniques, many genes were already mapped to the chromosome. This favoured status of knowing the genetic locus was due to the analysis of the members of the pedigree. On the other hand, the knowledge of genes present on the 22 autosomes and sex-chromosome; offers an entirely new set of challenges to the researchers. Nonetheless, pedigree analyses continued to be an invaluable tool for autosomal gene mapping till now. By co-segregating the phenotypes in pedigrees, researchers can generate linkage maps of the 22 human autosomes.

The interaction between the genes and the environment can establish the fact behind occurrence of most of the complex disorder. These interaction can help us to determine the gene affecting the trait. In case of Multifactorial disease, the linked genes can be determined by interactions between a gene or genes and the environment, that do not exhibit Mendelian ratios.For a graphic, interactive program to map quantitative trait loci by regression methods the software like **MAP MANAGER QT** can be used **.** The quantitative trait mapping can also be done via MAP MANAGER CLASSIC.The pure polygenic traits (i.e. no environmental influence) are infrequent. Moreover the polygenic traits are continuously varying and usually produce a range of phenotypes. Numerous methods can be used for investigating multifactorial traits for calculating RRR and FRR. This risk can be used to forecast recurrence of a multifactorial trait in a family. Also if the risk increases with severity of the trait, number of affected relatives, sex of family members, and increasing relatedness to an affected individual if known can help us in building the model . The Twin studies can be used to separate genetic and environmental contributions to a phenotype in case both members are identical (MZ) twins and not sibling.

Traditionally, the study of the role of family pedigree can provide answer to many questions like The pattern of inheritance of the disease, the risk of disease affecting the generations to come.The study of pedigree of families from diverse ethnic groups can reveal about the common disease symptoms,the pathway affected in the disease process ,genes associated

with the disease (in case more than one gene linkage studies can help us to determine part of chromosome associated with the disease), the relative risk of spread of the disease in the population.

The study of age of infected individuals and healthy individual can help us in identifying the onset age of the disease. The onset age and other such determinants can be used as important implications in order to delay the early onset of genetic disease.MENDEL , an online tool which can be used for segregation analysis, linkage calculations, genetic counseling, allele frequency estimation, and related kinds of problems.

The large-scale studies can help to isolate genes that contribute to complex diseases since individuals with alleles that predispose them towards a disease will statistically be more likely to be affected. Similarly, those with alleles that protect them from a disease will statistically be less likely to be affected. Many disease genes have been identified by looking for correlations between single nucleotide polymorphisms (SNPs) and complex diseases in large family and these studies are now being extended to population-SNP disease analysis. The association studies can be used for mapping Single nucleotide polymorphism (SNP) to identify multiple genes associated with a polygenic trait. These patterns can be studied among two populations to deviation in disease causing genes, susceptible genes.

A development in the field of diagnostics and genetic screening, DNA analysis can help in quick detecting of genetic disorders. The future of diagnostics and genetic screening is exciting and holds great promise.It should be extended upto the limit that diagnosis of the disease is possible prior to the birth of the offspring .The advances in techniques to study a members of the pedigree and their clinical analyse them will greatly expand the number of diagnosable conditions.The identification and counseling process for the patients having a high risk for a specific disorder would then be carried out in an appropriate manner.The advancements in the field will aid obstetrician with the knowledge of mode of inheritance of traits and then they will be able to indentify individual with high risk of developing the disorder.This information is nessecary for a practicing obstetrician ,since he is an interface between the patient and the scientist working in the areas of genetic research.