Chapter 6

Conclusion

The minor genetic variation among diverse ethnic groups predisposes them towards having a higher risk for certain genetic conditions than in the general population, such as cystic fibrosis (Northern European Caucasians), sickle cell disease (African Americans) and thalassemias (Mediterranean or Southeast Asian ancestry). Regardless of ethnic or racial background, each of us has five to 50 potentially disease-causing genetic variations. Most of the genetic changes in a population come and go pretty quickly, specifically mutations that reduce the fitness because the person dies and mutations are not generally passed to next generation. And if a new mutation is recessive, and does manage to stick, it would take millions of years, to see its effects. Not all mutations that are manifested are bad for an individual ,some can provide a benefit too, like the HBB (hemoglobin) gene mutation which causes sickle cell anemia led to the malarial tolerance seen in many people in inhabitants of Sub-Saharan Africa today. In recent years, geneticists and cancer researchers have focused attention on families with increased incidence of genetic diseases and various cancers. In many of these families, cancers such as breast cancer and colorectal cancer are common. A statistical analysis can reveal the insights factors causing the disease. But the basic necessity is to develop a pedigree. The question is, whether a single pedigree is enough. The answer to this question is no and therefore data-rich source are needed to be referred .The vast amount of data generated by researchers needs to be stored properly so that it can be communicated to large masses. The datasets grown, needs to be archived, curated, analyzed and interpreted. For this we require to full fill challenges like; having a convenient methods for proper storing, searching & retrieving necessary information. Thus creating and maintaining a databases means to handle large volume of data in a sustained manner.

A biological database is not just a collection of raw data in the form of facts and figure but an organized form of data, so that its contents can easily be accessed, managed, and updated according to the requirement. The database would be made available at http://dbpedigree.dce.edu website so that it can be made available to a multi-user system like medical professional, researcher's etc. Many of these disorders are genetic in origin and the increasing resolution of genomic technologies has enabled substantial increases in diagnosis rates in recent years. Nevertheless, most children with developmental disorders do not currently receive a genetic diagnosis. In such cases the advent of familial aggregation studies

raises the prospect of discovering many currently unknown disease genes and increasing diagnosis rates dramatically in the near future. However, our ability to discover millions of genetic variants in every genome is running far ahead of our ability to accurately identify individual disease-causing variants. Many of the online tools are available for KINDRED; PEDHUNTER, PEDIGRAPH etc. This interpretation gap is the fundamental challenge in human genetics today can only be resolved if the laboratory analysis can be carried out to come up with new discovery to solve the issues. The database and the webpage has been linked using PHP script. The dbPedigree database has 2850 pedigree entries covering 360 diseases, 450 genes, 820 mutations and 450 SNP's. In all 2850 pedigrees have been reported from Europe, America, Asia and islands also. This database can be accessed freely.