Abstract

Background:

Pedigree analysis, in its broadest sense is the process of drawing logical implications about a particular pedigree or set of pedigrees on the basis of familial aggregation evidences. The construction of an accurate family pedigree is a prerequisite for clinical genetic services and serves as an informational framework for human genetic research. It can also influence genetic studies and help in the selection of the most appropriate form of genetic analysis, that is important for counselling of affected member of a family. Additionally, identifying associated gene, mapping of its locus and identifying mutation also requires specification of the mode of inheritance of a disorder. The examination of a family pedigree aids the doctor in establishing the pattern of inheritance, and thereby assists them in identifying risk probability penetrance of gene expression in future generation. Thus it acts as a reference for social and biological relationships making clinician observant regarding the issues of blended families, adoption, deaths, pregnancy termination, and pregnancies conceived by assisted reproductive technologies. Correct interpretation of family pedigrees is essential for human genetic research and is particularly challenging when research teams collaborate to study families from different parts of the world having same genetic manifestations. Pedigree analysis also facilitates the identification of disorders where genetic mechanisms such as anticipation, mitochondrial inheritance, X-linked or dominant homozygous lethality are involved, along with the information regarding and differential age of onset of the disorder ,or the sex of the transmitting individual. Besides, scientists are publishing new data at a very fast pace. In order to make meaningful connections among worldwide scientific discoveries, there is a need for development of a database of pedigrees associated with genetic diseases.

Description:

The dbPedigree (Database of Pedigree associated with genetic defects); a relational database, contain; a collection genetic defect, its causable gene, associated mutations and related pedigrees. These pedigrees have been excavated from literature after an extensive mining process. The database also states the mode of inheritance of the disorder , the ethnicity of the proband as reported in literature. Also chromosomal location of the associated gene, along with the mutation (in the form of nucleotide change, deletion, inversion, insertion

etc.), SNP ID (have been reported from dbSNP). The citation reporting the pedigree has also been included in the records. The pedigree has been reported in the form of hyperlinks of the web link containing the image. It is a manually curated database reporting 360 diseases found to be associated with 450 genes .These many genes containing 1170 mutation out of which 450 are SNP's. In all 2850 pedigrees have been reported. This database is available at http://dbpedigree.dce.edu.