Genetic disorders are caused due to the presence of mutation in the DNA sequence of single gene or multiple genes of an individual. Most of these genetic disorders are heritable, which follow a specific pattern of inheritance. Several genetic markers have been identified to unravel the complexity of the disorder. Genetic markers are the variants in the DNA, which confer high level of probability of disease. There are different types of DNA markers namely, microsatellites, indels, Single Nucleotide Polymorphisms (SNPs) and Copy Number Variations (CNVs) used to identify the underlying genetic cause. The association of genetic markers with human disease has led to the identification of genes and genetic mutations responsible for several heritable diseases. These markers may have functional consequences by altering the function of the gene. Advances in molecular technologies have helped in the rapid identification of these markers.

Among these markers, SNPs have been extensively studied and are correlated with specific phenotype. Screening of SNPs in large number of individuals enables the prediction of susceptibility to a wide range of diseases. Thus, using SNPs effectively as genetic markers and identifying groups of markers traveling together in populations is a promising approach to identify the cause of the disorder and is also helpful in designing diagnostic tests and therapeutic treatments. Research on DD, provided information on candidate genes and associated SNPs and these SNPs were replicated in different populations through association studies.
In view of this, the present investigation was carried to further understand the genetic causes of DD in India. The findings and implications of the study are compiled and presented as follows:

**Section I** Reviews the literature of DD on epidemiology, family history, patterns of inheritance, genetic markers and candidate genes.

**Section II** Presents the identifications and analysis of family history of DD, consanguinity, inheritance patterns and SNPs associated with sub phenotypes of DD. These factors are investigated and discussed in the subsections - introduction, material and methods, observations and discussion.

**Section III** Summarizes the findings and brings the future prospects of these investigations.

**Section IV** Deals with literature cited in this thesis.

**Section V** Includes the list of publications made from this work and conferences attended.