PREFACE

Human diseases and sufferings are as old as humanity. A class of diseases that not only causes physical misery but also psychological trauma to the patients and to his family is genetic disorder effecting successive generation. (Snustad and Simmons, 2006). The problems of genetic disorders are enormous, millions of people are suffering from inherited diseases. One sixth of the world population is suffering from genetic disorders. The distressing and disturbing truth in this sad saga is that there is no cure available to these diseases and hence, the prevention of the inheritance of such diseases becomes absolutely essential.

Though human cytogenetics had started long back it was called as the Dark Age or pre hypotonic age until 1950s. Medical genetics came into existence in 1960s of the last century with the discovery of correct number of chromosomes and finding of the abnormal chromosomes in clinical syndromes. The molecular genetics had started in 1970s, the boon to classical human cytogenetics. From the last 47 years, after the report of extra 21st chromosome in Down syndrome, extra X chromosomes in Klinefelter and absence of X chromosome in Turner syndrome, tremendous work had been done throughout the world to elucidate the risk factors of these syndromes. One of the important questions being asked for several decades is that how the nondisjunction of autosomes and sex chromosomes causes a series of complex changes in individuals. Till now this remains as major challenge to be worked out.
It took 41 years of many researchers' efforts until the next historical step, which came on May 18, 2000, when the complete DNA sequence of human chromosome 21 was determined and published in Nature (Hattori et al., 2000). As human genetics enter into 21st century, researchers working in the field are optimistic that they may be on the brink of curing some of the common genetic disorders.

India represents one of the largest human diversity. In Indian context variability in human population is enormous due to inbreeding in different cultural groups, caste and sub caste system. Thus, in the present investigation, an attempt has been made to find out the etiological factors involved in Down syndrome and sex chromosomal aneuploidy. The findings and the implications of these studies are complied in the following sections.

Section I presents what is already known about genetic basis of common pediatric disorders in general and Down syndrome and sex chromosomal aneuploidy in particular.

Section II provides the prevalence, etiology and demographic factors of Down syndrome and sex chromosomal aneuploidy in Mysore population.

Section III summarizes the findings and brings out future prospects of these investigations.

Section IV records the literature cited in this thesis and

Section V deals with the list of publications made out of this investigation.