CHAPTER-I : INTRODUCTION

INTRODUCTION

The application of cytogenetic studies to clinical problems has proven to be a useful diagnostic tool and has enabled us to understand the pathogenesis of previously obscure conditions. Its contributions have been felt in the field of congenital anomalies including abnormal sexual and somatic development, mental retardation, infertility and spontaneous abortion. It has also helped in the study of the effects of certain environmental agents such as ionizing radiation, drugs and viruses. Thus, medical cytogenetics have become an integral part of the theory and practice of medicine.

Cytology, the study of cell structures, dates back to 1665, when Robert Hooke, first saw the cells of cork. The statement "omnis cellula and cellule" which is attributed to the German Pathologist, Virchow implies that all cells arise from pre-existing cells.

The cell consists of two main parts : the cytoplasm and the nucleus. In higher organisms, the cytoplasm contains the cell organelles which include mitochondria, microsomes, lysosomes and centrosomes. The nucleus contains the genetic material which is organised into morphological units; the chromosomes. The study of the morphology and behaviour of the chromosomes is called cytogenetics. Medical cytogenetics is that specialized branch which deals with the chromosomal changes in relation to pathological conditions in man. The function of the body is controlled by the genes which are located in the chromosomes. Even a minute disturbance in genes or gross disturbance in chromosomes could cause functional disturbances. Each chromosome contains the "genetic map of an individual" or programming on analogical functioning, which is being

inherited by individuals, half each from both the parents. If an individual inherits a faulty genetic map from either of his parents, it leads to a variety of disorders like failure to grow physically or mentally. In case of structural abnormality, in adult life it develops into failure to conceive or to father of a child. Faulty genetic structure affects almost all the systems of the body and as a result serious diseases like cancer and others occur.

All these are learned from peas, muci, corn and flies. These findings were extrapolated to comprehend the complexities of human genetics. The narrow hereditary bridge between the parents and the offsprings is now well understood in terms of molecular information, where code has now been revealed to the inquiring mind.

Long before the insight into the microcytochemical composition of the basic blocks of inheritance was gained, scores of attempts were made to understand the microphysical configuration of these genetic materials.

Minor technical difficulties plagued the study of chromosomes in the early part of the century. Another milestone in the history of cytogenetics was reached in 1956 when the exact number of chromosome in human beings was established. Since that day, there is almost an explosion of research work in the extremely important branch of biology.

With the progress in the methodology of chromosomal study, significance and correlation of clinical problem and its association with the cytogenetic abnormalities were explored.