CHAPTER-II : OBJECTIVES

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The cytogenetics has become an integral part of theory and practice of medicine in developed countries, but it is still an academic luxury in a developing country like ours.

The cytogenetics is a multidisciplinery branch where contributions are obtained from Paediatricians, Psychiatrists, Obstetricians, Physicians, Pathologists, Anatomists, Biochemists, Anthropologists, Zoologists and Botanists.

Paediatricians and Psychiatrists are exposed to congenital anomalies and conditions like mental retardation, delayed milestones etc.

Obstetricians are exposed to primary amenorrhoea, infertility, repeated abortions and ambiguous genitalia, etc.

Physicians are exposed to different inherited diseases, hemophilia, Duchnne's muscular dystrophies, albinism, haematological malignancies and different carcinomas, etc.

Ophthalmologists are exposed to retinitis pigmentosa and retinoblastomas, etc.

All above diseases and others are suggesting chromosomal aberrations or single gene disorders, hence cytogenetic study in such cases can be a valuable diagnostic tool in the field of clinical diagnosis and genetic counselling.

The chromosomal anomalies either spontaneous or induced by radiation therapy, chemical etc, lead to an abnormal development of body and gonadal differentiation. A normal appearing child is found infertile at adulthood and acquires phenotypic features of Turner syndrome, Klinefelter syndrome, testicular feminization syndrome, gonadal dysgenesis, poly X syndrome, or ambiguous genitalia, hermaphroditism, intersex stage etc., the sex chromosomal complex may be abnormal or at variance with the clinical and social sex.

Chromosomal aberrations are much more frequent and varied. There are significant causes of birth defects and fetal loss. Sex chromosomal and autosomal aneuploidies differ in many respects. The frequency of sex chromosomal aneuploidy as a whole is higher than that of autosomal aneuploidies among live births (2.3/1000 births versus 1.4/1000 births; Sankaranarayana, 1982 Review).

The difference may be because of (i) high fetal wastage of autosomal trisomies and (ii) sex chromosomes may be more prone to meiotic errors than autosomes and hence one can find large number of sex chromosome disorders. In addition to sex chromosomal abnormalities hormonal factors are also responsible for some abnormal differentiation and development. These abnormalities may be of genotypic or phenotypic, affect the individual in many ways :

1. Social behaviour of individual, in case of XY females, XX males, true hermaphroditics, mosaics etc. is affected.

2. There is a confusion of sex in the individual with ambiguous genitalia.

3. Sex chromosomal and autosomal abnormalities might lead to infertility in males and to spontaneous abortions in females.

4. The behavioural patterns, intelligence, IQ, mental capacity, growth in both autosomal and sex chromosomal anomalies are altered, e.g. Down's syndrome (+21), Klinefelter syndrome (XXY), XYY and Turner syndrome (XO).

Survival of human race is possible only when there is a normal growth and sex development, differentiation and function. Individuals with gross physical and sexual anomalies cannot fit themselves properly in the society unless the condition is diagnosed at a very early age and managed in the best possible way.

This study was taken up for the challenge it offers, in view of the following objectives :

1. To find out the autosomal and sex chromosomal abnormalities in the sufferers for the subsequent management of the patients and to extend counselling to the parents for the risk of future pregnancies; for the welfare of not only the family but also the society.

2. To correlate genotypic and phenotypic features in both autosomal and sex chromosomal abnormalities.

3. To understand role of autosomes and X and Y sex chromosome in normal sex development.

4. To evaluate the chromosomal and other responsible for the somatic and sex disorder in the patient.