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UNDERSTANDING DIFFERENT FACETS OF CHROMOSOME ABNORMALITIES: A 18-YEAR CYTOGENETIC STUDY AND INDIAN PERSPECTIVES

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Worldwide; at least 7.6 million children are born annually with severe genetic or congenital malformations and among them 90% of these are born in mid and low income countries. Precise prevalence data are difficult to collect, especially in developing countries, owing to great diversity of conditions and also because many cases remain undiagnosed. The genetic and congenital disorder is the second most common cause of infant and childhood mortality and occurs with a prevalence of 25-60 per 1000 births. The higher prevalence of genetic diseases in a particular community may, however, be due to some social or cultural factors. Such factors include tradition of consanguineous marriage, which results in a higher rate of autosomal recessive conditions including congenital malformations, stillbirths, or mental retardation. Genetic diseases can vary in severity, from being fatal before birth to requiring continuous management; their onset covers all life stages from infancy to old age. Those presenting at birth are particularly burdensome, and may cause early death or life-long chronic morbidity. Genetic testing for several genetic diseases identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. Several hundred genetic tests are currently in use, and more are being developed. Chromosomal abnormalities are major cause of human suffering, which are implicated in mental retardation, congenital malformations, dysmorphic features, primary and secondary amenorrhea, reproductive wastage, infertility neoplastic diseases. Cytogenetic evaluation of patients is helpful in the counseling and management of affected individuals and families. We present here especially chromosomal abnormalities which form a major part of genetic disease burden in India. Different programmes on chromosome research and human reproductive genetics primarily relate to infertility since this is a major public health problem in our country, affecting 10-15% of couples. Prenatal diagnosis of chromosomal abnormalities in high-risk pregnancies helps in detecting chromosomally abnormal fetuses. Such couples are counseled regarding continuation of pregnancy. In addition to the basic research, the team is providing chromosome diagnostic services that include conventional and advanced techniques for identifying various genetic defects. Other than routine chromosome diagnosis for infertility, also include patients with short stature, hypogonadism, undescended testis, microcephaly, delayed developmental milestones, familial and isolated mental retardation, and cerebral palsy. Thus, chromosome diagnostics has found its applicability not only in disease prevention and management but also in guiding the clinicians in certain aspects of treatment. It would be appropriate to affirm that chromosomes are the images of life and they unequivocally mirror the states of human health. The importance of genetic counseling is increasing with the advancement in the field of genetics. The genetic counseling can help families to cope with emotional, psychological and medical consequences of genetic diseases.

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