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Book Review

Chromosomal Abnormalities and Genetic Counseling
by R.J. McKinlay Gardner and Grant R. Sutherland
577 Pages, Oxford Univ. Press Inc. (2004) 3rd Edition (Hard bound)

Through each successful edition, Gardner and Sutherland's "Chromosomal Abnormalities and Genetic Counseling" has proved to be outstanding text for genetic counseling and chromosomal abnormalities to diverse scientific community involved in chromosomal pathologies and implication thereof. Now the extensively revised 3rd edition of this book adds on the account of conceptual consolidation. With its well-organized data book provides ready access to the reference and text on the specific abnormalities. Also in this edition, incorporated new content on the uniparental disomy and disorders ensures the understating of some the chromosomal pathologies, where no much standard text available apart from the individual case histories.

The book starts with the section on basic concepts of medical cytogenetics and chromosomal pathologies. Section's three chapters on "Elements of medical cytogenetics", "The origin and consequences of chromosome pathology" and "Deriving and using a risk figure" expose the reader to this field in a very clear and balanced manner. The next section *Parent with chromosomal abnormality,* perhaps the largest and richest section the book, blanket twelve chapters. The first of this section on "Autosomal reciprocal translocation" deals with the usual form i.e. simple or two ways, reciprocal translocation. The special cases of translocations involving sex chromosomes and those of complex (Robertsonian) translocations are dealt in the next two chapters. Each chapter on translocation examines the biology and meiotic behavior involved in the respective type of translocations along with the mode of segregation and prediction of sergeant outcomes. The complexity and pathology involved has been discussed vis-à-vis to the risk factor assessments and genetic counseling. The text is supported with rich data, photographs and diagrams. The chapter seven of the book and forth of the section sets in for the understanding of certain very rare chromosomal pathologies involving centromere fission, complimentary isochromosomes and telomeric fusions. The next two chapters i.e. eight and nine deals with the phenomenon of inversion and insertion respectively. The chapters discuss the different forms of such structural rearrangement, consequences of it, and possible genetic counseling interface. The tenth chapter examines in detail an uncommon chromosomal rearrangement – the ring chromosome. The examples of rings of different chromosomes along with the consequences thereof have been given in the chapter. In the eleventh chapter chromosomal pathologies arising due to the complex rearrangement in the chromosomes are given. The pathologic outcomes and genetic counseling interventions of major sex chromosome abnormalities (the XXY Klinefelter syndrome, 45,X Turner syndrome, XXX, XYY) their mosaic variants and partial forms have been detailed in chapter twelve. The chapter thirteen explains the parental autosomal aneuploidy and resultant consequences. A full chapter (i.e. chapter fourteen) is dedicated to genetic understanding of fragile X syndrome. In chapter fifteen chromosomal variants that have no phenotypic consequences apart from mere structural variation in the chromosomes are discussed. The next section of the book "*Normal parents with a chromosomally abnormal child"* starts with its first chapter (chapter 16) on understanding the biology and phenotypic consequences of the major trisomies (13, 18, 21), sex chromosome aneuploidies, and polyploidy amongst the offspring's of karyotypically normal parents. The next chapter (chapter 17) is a good content on chromosomal structural rearrangements, mechanism of their formation, and phenotypic pathologies involved. The chapter eighteen focuses on the conditions of imperfect sex differentiation particularly the cases where the process has taken an extreme opposite path i.e. XY females and XX males. Emphasis is given on the cases where cytogenetic and molecular genetics comprise the key diagnostic investigations. In the chapter twenty the individual cases of the phenomenon of uniparental disomy are

put forth in the form of a standard text. The different forms of uniparental disomy have been discussed in detail along with the consequent phenotype with respect to each human chromosome. Some of the most common imprinting disorders and their causative uniparental disomy along with the other resultant pathologic conditions are also detailed in this chapter. The section *"Reproductive Failure"* discusses the intricacy and frequencies of the chromosomal abnormality at each stage of reproductive process. It shows the vulnerability of process of conception and pregnancy as well the robust normal selection. The section *"Prenatal Diagnosis"* is divided into four chapters. The first one (chapter 22) examines the risk of fetal trisomies with respect to parental age and screening methodologies for such trisomies. Chapter 23 discusses the prenatal diagnosis procedures which are perhaps is the major application tool in medical cytogenetics. The diagnostic strategies of each procedure are detailed in view of the available data. A detailed experimental approach has also been given. A separate chapter (chapter 24) is dedicated to the new field of preimpalntation genetic diagnosis. This chapter outlines the target group of such procedures and diagnosis, clinical and laboratory procedures involved and genetic counseling strategies. The chapter 25 of the book deals with the role of a genetic counselor in assessment of phenotype and decision making after the prenatal diagnosis. Facts and figures of Mosaicism and other chromosomal aberrations in prenatal diagnostic assessment have been discussed in detail. The last chapter (twenty six) of the book reviews the effect of therapeutic and non-therapeutic (environmental) agent that could conceivably disturb the chromosomal distribution at gametogenesis.

The book provides the knowledge of cytogenetics fundamental to clinical practice. It introduces readers to the concepts, principles and practices of genetic counseling through analysis of genetic risk assessment, and counseling frameworks involved in various chromosomal abnormalities. The book also provides an overview of medical genetics spanning the prenatal, pediatric and adult disciplines.

Satish Kumar Yadav
Anthropological Survey of India
Mysore, Karnataka, India