

Book reviews

Human Cytogenetics: Constitutional Analysis

Edited by DE Rooney

Oxford University Press, New York; 2001. 282 pp. £32.50, paperback. ISBN 0–19–963839–X

Human Cytogenetics: Malignancy and Acquired Abnormalities

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Cytogenetics has traditionally been the realm of specially trained and very experienced scientists. However the advent of new techniques, particularly those improving visualisation of abnormalities, have opened this field up to a growing number of researchers from varying backgrounds, including those who lack the technical training of cytogenetics. All cytogeneticists, and especially researchers and trainees, would benefit from the expert general knowledge of cytogenetics that is imparted via these books. Each is an updated (3rd) edition of a widely used previous publication in the Practical Approach Series.

Human Cytogenetics: Constitutional Analysis opens with a good introduction to different types of chromosome abnormalities – how to recognise and describe them (according to ISCN), how they arise and subsequent effects they may have on cells or individuals. This is followed by a detailed description of basic lymphocyte culture techniques and metaphase preparation. The purpose or effect of reagents at different stages is discussed, as are various other factors which can affect the quality of preparations. An overview is given of the most common and widely used staining techniques, including the biochemical basis of their action, the cytogenetic features targeted, and different banding patterns. Consideration of sterility/contamination, choice of glassware/plasticware, incubators, growth conditions, and media for cultures is raised in a chapter about the cytogenetics of pregnancy. Sampling procedures for chorionic villus, amniotic fluid, solid tissue, sperm, oocytes and embryos are described, including associated maternal contamination and the types of cultures derived. Detailed guidance of cytogenetic analysis is provided, ie what to look for and which tests will be most appropriate, on the basis of clinical features (age at presentation, phenotypes and family history). Much emphasis is placed on correctly and carefully interpreting results and preparing reports.

Basic FISH is well described with the purpose and

potential problems of each stage (and reagents) discussed, from probe choice and labelling to microscopy and interpretation of results. Helpful troubleshooting suggestions are given. The particular delicacies of human meiotic studies, in contrast to mitotic studies, are conveyed, with sample acquisition and preparation, chromosome analysis, and different meiotic stages described. To conclude, the extremely important issue of microscopy and image analysis is considered. This excellent chapter should ensure that all readers can properly understand their microscope and its imaging capabilities.

Human Cytogenetics: Malignancy and Acquired Abnormalities begins with an introduction to a broad range of bone marrow and blood culture techniques, from sample collection and cell counting to long-term preservation. Potential traps in analyses are identified, and molecular techniques introduced. This is followed by a sequence of chapters describing myeloid leukaemias, acute lymphoblastic leukaemias, and lymphomas and chronic lymphoid leukaemias. For each, their classification, and clinical and cytogenetic features are discussed. Acquisition and appropriate culturing of samples is touched upon, and associations with specific chromosome rearrangements, deletions, and molecular and numerical abnormalities are detailed. To end this series, a general discussion of haematological neoplasms considers clonality and the contribution of cytogenetics to the understanding of neoplasia development and improved diagnoses. The remainder of this text discusses the use of newer and more complicated FISH techniques including M-FISH/SKY, subtelomeric probes, interphase and RNA FISH, and CGH, and their diagnostic practicalities. Solid tumour sampling, culturing, karyotyping and aberrations follow, as do techniques for scoring structural aberrations, sister chromatid exchange, micronuclei, and radiation dosimetry in response to *in vivo* mutagen-induced chromosome damage. Lastly, the features of numerous chromosome instability syndromes are delineated.

The only disappointing thing about these books is the quality of some of the figures, which are better in the previous editions and similar publications, and noticeable in a field which appreciates exceptional images. However the high quality of the written content more than makes up for this. There is some redundancy in the topics covered, but due to their context this generally results in additional understanding and what repetitiveness does occur serves to re-emphasize important points. Relationships between clinical and cytogenetic features, and prognosis, and their relative importance, are defined for all types of disease. Similarly, appropriate exchange of information between different health professionals, including clinicians, haematologists, cytogeneticists, researchers, and laboratories is raised.

Throughout these books the relative advantages, disadvantages and limitations of every technique are raised and suggestions made for choosing between them. New

and emerging techniques are mentioned with enthusiasm. Clear protocols are provided in sufficient detail to be attempted without additional research, and are easily distinguishable from the main text. The fickle nature of chromosomes and their preparation does mean, however, that many cytogeneticists develop their own slight, but preferred, variations to these protocols.

Human Cytogenetics: Constitutional Analysis is perhaps the more useful of the two, largely because it encompasses all types of cytogenetic abnormalities that

may arise in any tissue. However, given the incidence and increasing cytogenetic understanding of cancer, both books would be invaluable to any cytogenetics laboratory, wherever their interest lies.

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