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Molecular Cytogenetics

Protocols and Applications

Edited by

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Preface

The new techniques of molecular cytogenetics, mainly fluorescence *in situ* hybridization (FISH) of DNA probes to metaphase chromosomes or interphase nuclei, have been developed in the past two decades. Many FISH techniques have been implemented for diagnostic services, whereas some others are mainly used for investigational purposes. Several hundreds of FISH probes and hybridization kits are now commercially available, and the list is growing rapidly. FISH has been widely used as a powerful diagnostic tool in many areas of medicine including pediatrics, medical genetics, maternal–fetal medicine, reproductive medicine, pathology, hematology, and oncology.

Frequently, a physician may be puzzled by the variety of FISH techniques and wonder what test to order. It is not uncommon that a sample is referred to a laboratory for FISH without indicating a specific test. On the other hand, a cytogeneticist or a technologist in a laboratory needs, from case to case, to determine which procedure to perform and which probe to use for an informative result. To obtain the best results, one must use the right DNA probes and have reliable protocols and measures of quality assurance in place. Also, one must have sufficient knowledge in both traditional and molecular cytogenetics, as well as the particular areas of medicine for which the test is used in order to appropriately interpret the FISH results, and to correlate them with clinical diagnosis, treatment, and prognosis.

Molecular Cytogenetics: Protocols and Applications provides reliable protocols for most of the FISH techniques in a step-by-step and easy-to-follow style for laboratory physicians and scientists who offer diagnostic services in genetics and oncology. Notes for best results and troubleshooting are offered based on the authors' first-hand experience. In many chapters, the authors have provided an extensive review on the applications of the technology and its sensitivity, limits, and pitfalls. Several review chapters on some particular topics have also been included. Although the major focus is on diagnosis, the state-of-the-art protocols detailed in depth here can be very useful for all scientists who are interested in genomic research in the areas of human development and the molecular biology of human cancer.

It is hoped that *Molecular Cytogenetics: Protocols and Applications* would serve as a major source of guidance and reference for the providers of

diagnostic services, including cytogeneticists, pathologists, technologists, trainees, and students in cytogenetics, hematology, and molecular pathology. The authors have written here with particular considerations for the needs of clinicians, as well as other health care professionals, who utilize molecular cytogenetic tests for the diagnosis and clinical management of patients with developmental disorders, reproductive problems, or oncologic diseases.

Yao-Shan Fan

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