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FLUORESCENT *IN SITU* HYBRIDIZATION (FISH): CURRENT STATUS IN CLINICAL CYTOGENETIC DIAGNOSTICS.

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Abstract

FISH is a powerful technique for detecting DNA or RNA sequences in cells, tissues and tumors. This molecular cytogenetic technique enables the localization of specific DNA sequences with in interphase chromatin and metaphase chromosomes and the identification of both structural and numerical chromosome changes. FISH uses fluorescent DNA probes to target specific chromosomal locations within the nucleus, resulting in colored signals that can be detected using a fluorescent microscope. Compared to the conventional cytogenetic (CC), metaphase karyotype analysis, FISH does not require cell culturing, and can directly use fresh or paraffin embedded interphase nuclei for a rapid evaluation. With the discovery of numerous disease-related genes in recent years, the application of FISH broadened to include more genetic diseases, hematologic malignancies and solid tumors. FISH detection of BCR/ABL1 translocation, HER2 amplification and ALK rearrangement is critical for guiding targeted therapy in chronic myeloid leukemia, breast cancer and lung adenocarcinoma, respectively. FISH tests have been recognized as vital components of personalized medicine.

Keywords: FISH, Cytogenetics, Clinical diagnostics

Introduction

Recurring chromosomal abnormalities are associated with distinct subtypes of leukemia or lymphoma with unique morphologic, immunophenotypic, and clinical features such as response to therapy (Le Beau, 1993 and Mitelman, 1994). Thus, cytogenic analysis of an individual's malignant cells plays a major role in the diagnosis and subclassification of a hematologic neoplasm. Molecular analysis has revealed that recurring chromosomal abnormalities results in the altered function of oncogenes, thus cytogenetic aberrations represent genetic mutations that are involved in the pathogenesis and progression of human tumors (Look, 1997). The development of molecular hybridization techniques such as fluorescent in situ hybridization (FISH) has a major impact on efforts to detect and characterize the genetic changes that give rise to human tumors. With probes designed to identify specific chromosomes and chromosomal regions, FISH is a cytogenetic technique used to detect and localize the presence and absence of specific DNA sequence on chromosomes. This method permitted investigators to expand the assayable target, to allow whole chromosome painting procedures (Lichter et al., 1988). Chromosome analysis by FISH have led to marked progress in cytogenetics research (Trask, 2002). FISH is used routinely by cytogenetics and pathology laboratories to identify recurring chromosomal abnormalities (Table 1). The combination of cytogenetics, FISH and molecular analysis provides a powerful tool for diagnosing and sub classifying malignant disease into clinically and biologically relevant subgroups (Gozzetti and Beau, 2000).