ABSTRACT

DETECTION OF GENETIC OR MOLECULAR ABERRATIONS ASSOCIATED WITH CANCER

Systems, apparatus, and methods are provided for determining genetic or molecular aberrations in a biological sample from an organism. Biological samples including cell-free DNA fragments are analyzed to identify imbalances in chromosomal regions, e.g., due to deletions and/or amplifications in a tumor. Multiple loci are used for each chromosomal region. Such imbalances can then be used to diagnose (screen) a patient for cancer, as well as prognosticate a patient with cancer, or to detect the presence or to monitor the progress of a premalignant condition in a patient. The severity of an imbalance as well as the number of regions exhibiting an imbalance can be used. A systematic analysis of non-overlapping segments of a genome can provide a general screening tool for a sample. Additionally, a patient can be tested over time to track severity of each of one or more chromosomal regions and a number of chromosomal regions to enable screening and prognosticating, as well as monitoring of progress (e.g. after treatment).

[Figure 10]