Molecular Karyotyping

A leap in cancer diagnosis

we see what others don’t
Molecular Karyotyping: Beyond Conventional Cytogenetics

- Cytogenetic analysis provides valuable diagnostic and prognostic information for the evaluation of hematologic malignancies. Traditional and new techniques have been applied in clinical oncology with a variable range of resolution and sensitivity, including chromosome analysis and fluorescence in situ hybridization (FISH).
- However, the resolution of these techniques is still low since the detection of unbalanced rearrangements is not well defined. In addition, chromosome analysis cannot be performed when the culture fails.
- Recently, the application of DNA array-based technologies such as array comparative genomic hybridization (aCGH) has enabled the detection of previously undetected copy number changes (CNCs) and the more precise determination of genomic breakpoints of regions that are gained or lost with an increased resolution and sensitivity.

GenArray™ is a custom-design microarray that contains 64,000 DNA oligonucleotide probes spaced at approximately 50Kb intervals across the genome. There is a higher probe density in cancer-relevant regions, resulting in higher sensitivity and greater resolution allowing for the detection of even the smallest genomic variations with clinical relevance.

Gains or losses greater than 1Mb and observed in at least 30%–40% of the patient DNA sample are reported.

Figure 1: A molecular karyotype showing gains and losses across the genome. Gains are depicted in green and losses in red.

- Array CGH technology does not rely on metaphase chromosomes, and, therefore, the DNA sample can be obtained from cells in any stage of growth.
- Use of oligonucleotide array allows greater coverage and probes may be present every 21Kb, as opposed to Bacterial Artificial Chromosome (BAC) arrays, which have a 1Mb–2Mb coverage.

This technology should be used in conjunction with cytogenetic analysis, as it cannot detect balanced translocations and inversions.

For further information, please contact us at 1 800 627 1479 or visit our website, www.genpathdiagnostics.com.
Clinical Indications

**Myeloproliferative Neoplasms:** In a number of cases, patients with Polycythemia Vera (PV) and Essential Thromobocythemia (ET) show normal cytogenetics even though array CGH analysis can detect a number of chromosomal aberrations.

**Lymphoproliferative Disorders:** Lymphoid malignancies and plasma cell disorders that involve end-stage cells are difficult to analyze using cytogenetics.

**Myelodysplastic Syndrome (MDS):** Normal karyotypes in chromosome-banding analysis represent 40%–45% of MDS and Acute Myeloid Leukemia (AML) patients. GenArray™ currently facilitates the genetic diagnosis of patients with MDS where chromosome analysis was not successful due to the nature of the disease (culture failures, low resolution karyotypes or normal results). MDS patients are at high risk of developing AML, and it is important to screen these patients for any chromosomal abnormalities that cannot be detected by standard cytogenetics.

Array CGH also has proven utility in Chronic Lymphocytic Leukemia (CLL) and other leukemias.

Figure 2: A GenArray™ result showing a 2.9MB deletion on chromosome 4.

References:


### Ordering Information

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<th>Test Name</th>
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