

Title: "A complete platform for the analysis of copy number changes in clinical cytogenetics"

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Abstract:

aCGH is a powerful technique that has revolutionized the analysis of chromosomal aberrations in the new millennium.

PerkinElmer has developed a complete platform for aCGH including microarrays, reagents for labeling and hybridization, scanners and analysis softwares. Each and every component of our platform play an important role in generating a clinical report.

We propose 2 BAC-arrays for aCGH. The first -Constitutional Chip 3.0- is a targeted microarray for detecting syndromes with known genotype-phenotype correlation. The second one -Constitutional Chip 4.0- is a whole genome array with overall resolution of 590 Kb. This array investigate subtelomeric regions at a median resolution of 250 Kb, examine more than 90 known genetic conditions and explore at a high resolution (250Kb) the X chromosome for the analysis of X-linked mental retardation cases.

PerkinElmer has developed and patented the cyanine dyes (Cy3 and Cy5) that are included in the labeling kit.

We have recently developed a high resolution scanner (max resolution 3 microns), with simultaneous dynamic autofocus scanning. This scanner is extremely fast and can scan a whole slide at 5 microns in 4 minutes.

Our new analysis software is totally interactive. It starts from loading the tiff images from the scanner, performs automatic grid placement, quantifies the fluorescent spots with a proprietary autosegmentation method and visualizes the normalized data. The generated data are totally interactive with the related images and with the raw data, allowing the user to analyze the consistency of the data with the original fluorescent spots. The software performs also quality controls on the overall performance of the array, such as standard deviation of each chromosome or of all autosomes, number of responsive clones, S/N ratio, excluded clones, and then generates a report with all these informations and with the detected gains and losses in ISCN nomenclature.

We also provide the BAC-DNA for FISH confirmation of the results.

This complete platform can be used in clinical settings for the analysis of chromosome aberrations in pre-natal, post-natal, preimplantation and oncology: examples of aCGH results generated with different kind of samples will be shown in the presentation.