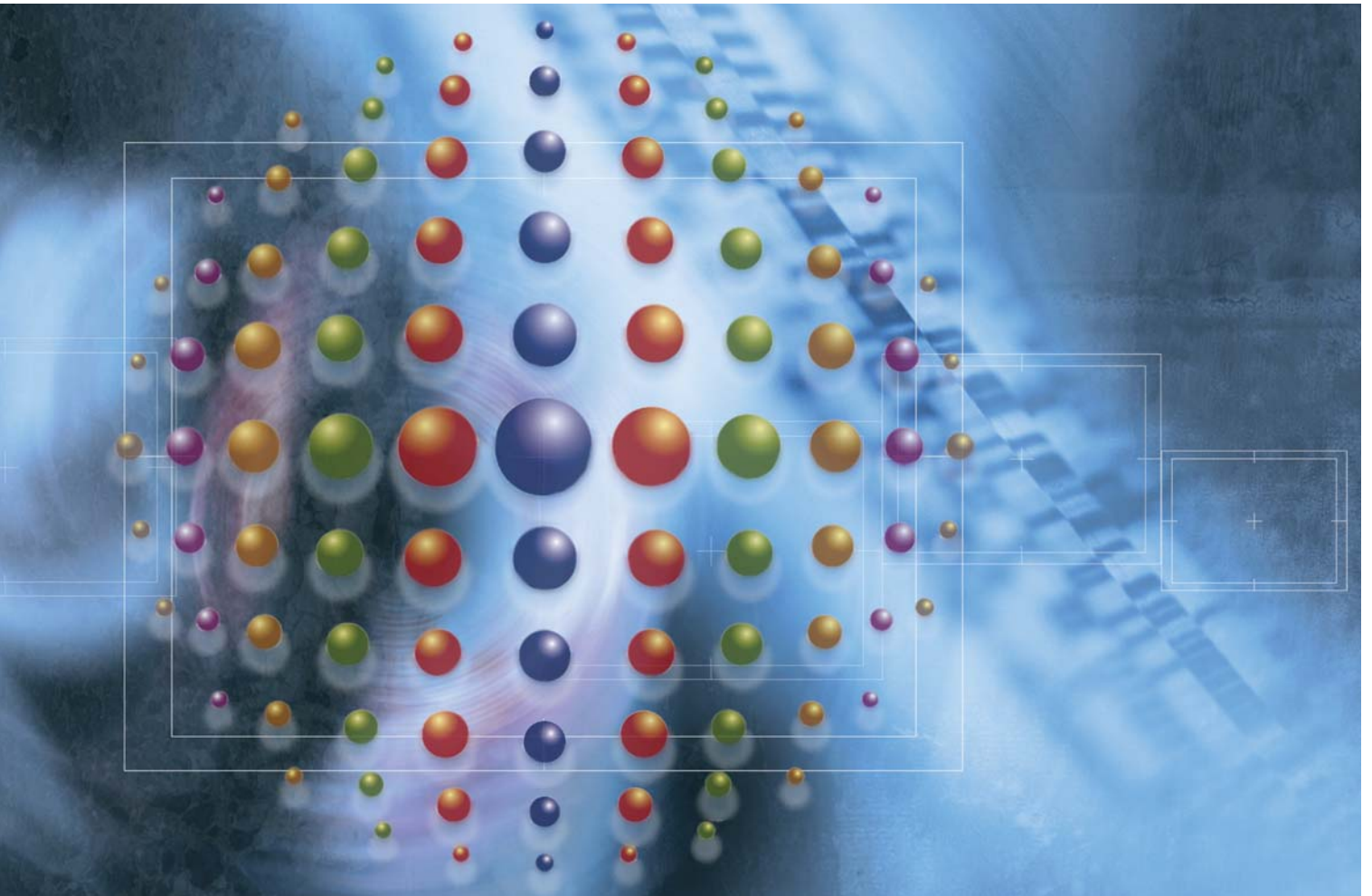


Driving the Revolution in Cytogenetics



High throughput, high resolution
molecular karyotyping

Objective and fast identification of ch



Array Comparative Genomic Hybridization (aCGH) provides genome-wide analysis of chromosome copy number and structural change at high resolution, generating more objective and useful information from a single analysis. aCGH is fundamentally changing the way genetic information is derived from chromosomes.

Chromosomal abnormalities

aCGH provides major advantages over existing methods. It removes the subjectivity of classical karyotyping, and it provides breadth of coverage far superior to FISH. A single aCGH test will replace multiple, iterative FISH assays, and allow you to obtain high-resolution, genome-wide analyses with major savings of work and time.

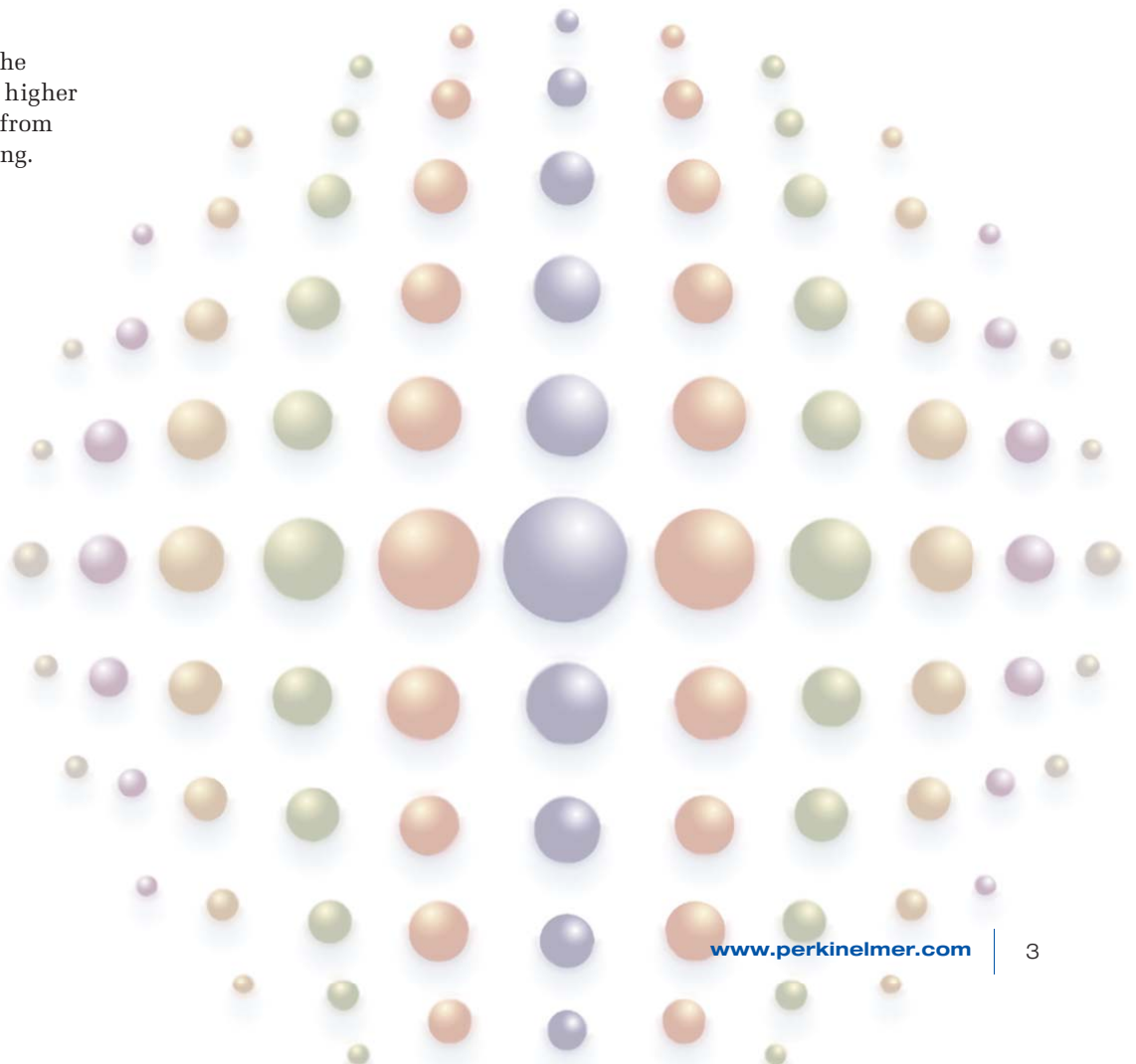
Conventional cytogenetic testing is time consuming, laborious, and requires expert technical data analysis to ensure a reliable result. aCGH does not require living cells, dramatically reducing the time required to deliver results on multiple loci from a single sample. Automated analysis produces objective results and provides tools to support cytogenetic interpretation.

aCGH simultaneously assays the genome at selected loci with a higher resolution than that available from conventional cytogenetic testing.

aCGH provides major benefits in the investigation of the genetic causes of mental retardation and developmental delay

aCGH provides a superior alternative to telomere FISH for the investigation of genomic errors associated with developmental disabilities and mental retardation. aCGH also easily detects duplications of common microdeletion regions which will not be detected by metaphase FISH. Taken together these factors eliminate the need to perform loci specific tests for individual constitutional conditions as seen with conventional cytogenetic technologies.

- **High resolution karyotyping**
- **Comparable to hundreds of simultaneous FISH assays**
- **Results within two working days**
- **Automated data analysis for speed and objectivity**



Reliable results through quality controlled reagents and proprietary chip design

The Constitutional Chip kit includes two microarrays with more than 600 BAC clones. Each clone has been end-sequenced and its cytogenetic position identified by pairwise comparison using public databases followed by both in-house and independent verification. All clones are printed in triplicate and analysis performed through dye-swap on two arrays. Together with the validated and quality controlled reagents for performing both hybridization and labelling, and an optimized protocol the Constitutional Chip kit brings exceptional reliability to molecular karyotyping.

The proprietary targeted design of Constitutional Chip version 3.0 simultaneously evaluates trisomies, 41 recognized microdeletion or microduplication syndromes plus 41 unique subtelomeric regions. All loci are carefully chosen based on clinical significance and association to known phenotypes. Comprehensive coverage is ensured by a minimum of three clones for each targeted region.

Proprietary design provides absolute confidence in results

- **Comprehensive coverage of clinically significant loci**
- **6 data points analyzed per clone**
- **Control and calibration clones for data normalization**
- **Optimized protocol and quality controlled reagents**
- **Automated data analysis with ScanArray® Gx PLUS and SpectralWare®**

Clear highlighting of abnormalities

After running the Constitutional Chip kit, the resulting pattern of differentially fluorescing spots can be scanned with the PerkinElmer ScanArray® Gx PLUS scanner. Together with the SpectralWare™ software a detailed structural analysis of chromosome regions is provided. Operation is simple and fully automated, saving time, removing operator error and subjectivity and ensuring experimental reproducibility. Grid alignment and signal estimation are followed by normalization to remove bias and exclusion rules to ensure that only reliable results are returned. Replicates are combined and regions of chromosomal abnormality are identified.

aCGH for Constitutional Molecular



Run it in *Your* lab
and avoid the need
to send samples out
for testing

Karyotyping

aCGH for Constitutional Molecular Karyotyping

Array CGH provides high-resolution measurements of chromosomal abnormalities. Immobilized BAC probes interrogate a labeled genomic DNA sample with the same hybridization chemistry as FISH. The sample is labeled with one fluorophore and a normal reference is labeled with a second fluorophore. The ratio of the two fluorophores at each array element corresponds to genomic amplification or loss at the chromosomal locus of that probe. All known trisomies, 41 microdeletion and microduplication syndromes and 41 subtelomere regions are measured simultaneously.

PerkinElmer Constitutional Chip®*

Robust

- “Dye-swap” minimizes bias - two independent arrays with reciprocal labeling reactions on the same chip
- All BAC probes are printed in triplicate on/in each array
- ScanArray® and SpectralWare® automatically normalize signals and calculate gains and losses

Comprehensive Coverage

- Able to detect all known trisomies
- > 600 BAC probes covering 41 clinically significant regions
- 41 subtelomeric regions

High Quality

- All BAC probes are end-sequenced
- Stringent quality control of each printing batch
- All arrays go through 100% visual inspected
- Quality controlled labeling, hybridization, and wash reagents
- Optimized protocol

Global Scale and Support

- PerkinElmer supplies a complete aCGH solution: microarrays, labeling, hybridization and wash reagents, ScanArray® microarray scanners and image processing software, and SpectralWare® array CGH analysis software

Constitutional Chip® aCGH

- Full spectrum analysis of developmental disorder loci
- Simultaneous detection of more than 600 BAC probes
- Objective numerical results
- Automated scanning and analysis

FISH

- FISH will only provide information about the probe being tested, other aberrations will not be detected
- Restricted multiplexing possibility
- Limited number of commercial probes available
- Needs specialised camera and image capture system

* For Research Use Only

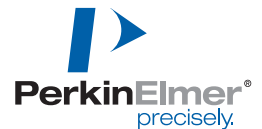
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