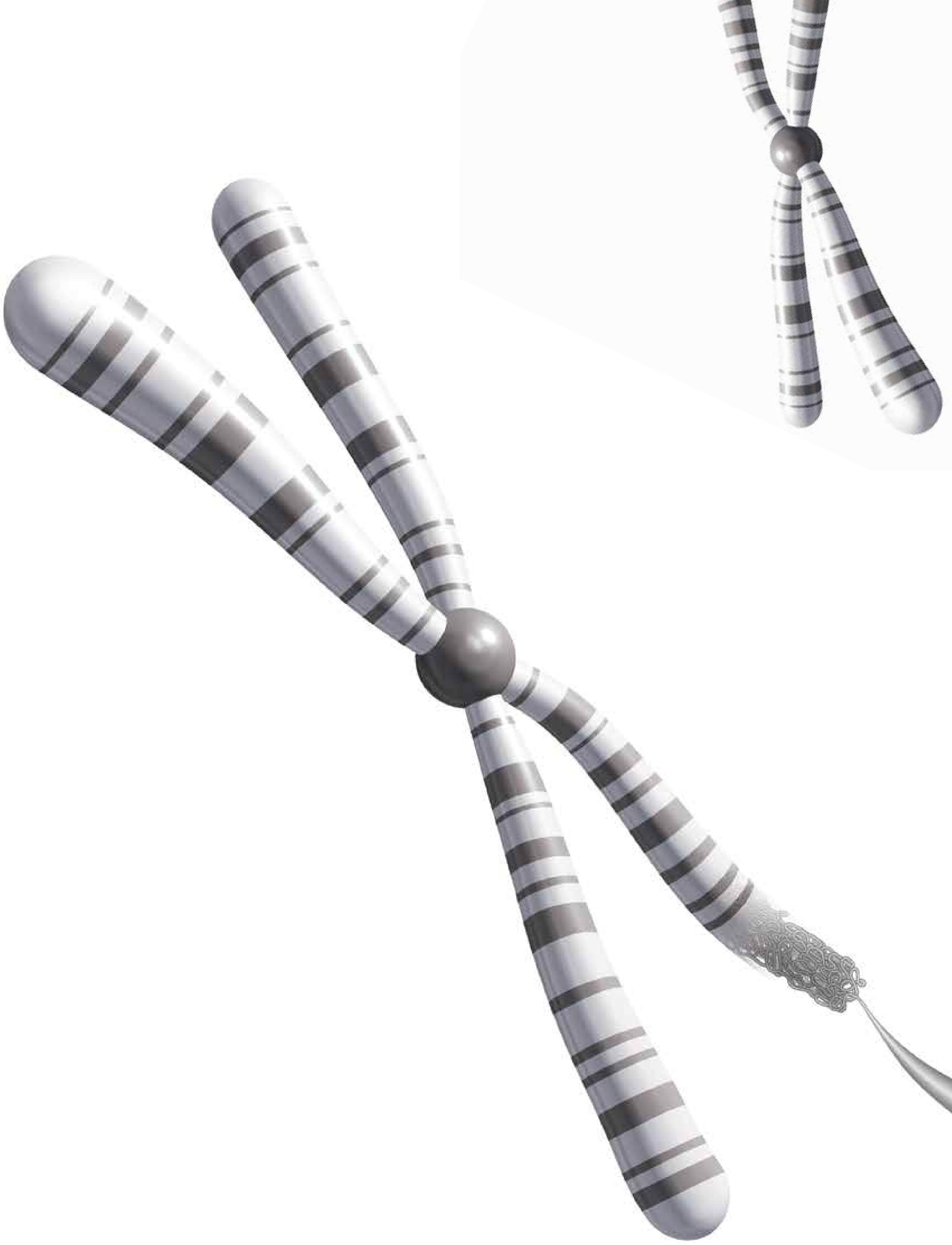
Getting high-quality cytogenetic data is a SNP.

illumina®



SNP data. Increased insight.

Cytogenetics is at the forefront of the study of cancer and congenital disorders. And we put you at the forefront of cytogenetics.

Illumina SNP array technology combines genotype and intensity information to detect chromosomal aberrations. This gives you the ability to identify chromosomal abnormalities that other arrays can’t recognize.

See the data from every angle. Get better data quality and reproducibility. Simplify your workflow with automation options.

Get better results faster.
Add SNPs to your data.

- To learn more, visit www.illumina.com

More information. Superior detection.

Informative markers. High resolution. By combining the genotype and intensity information provided by Infinium® BeadChips, you can profile both copy number variation and copy-neutral events, and also identify important factors such as mosaicism that can elude standard cytogenetic analysis.

Get complete coverage with meticulously selected SNPs and probes for detection of:

- Amplifications and deletions
- Loss of heterozygosity (LOH)
- Uniparental disomy (UPD)
- Mitotic recombination
- Gene conversion events
- Unbalanced translocations

Infinium BeadChips are suited to most cytogenetics applications, including studies of developmental disorders and cancer. With broad genomic coverage and specific targeting of important known regions, you don't have to choose between focused and broad arrays.

Intelligent Marker Selection

SNP genotyping markers are preferred because they provide greater information and are more robust for finding cytogenetic aberrations. The SNP markers on Infinium BeadChips are supplemented with intensity-only non-polymorphic probes to ensure a high-density backbone and broader coverage of high-value regions. SNP marker information generates higher signal-to-noise ratios for copy number calculations., enabling detection and identification of even minor events.

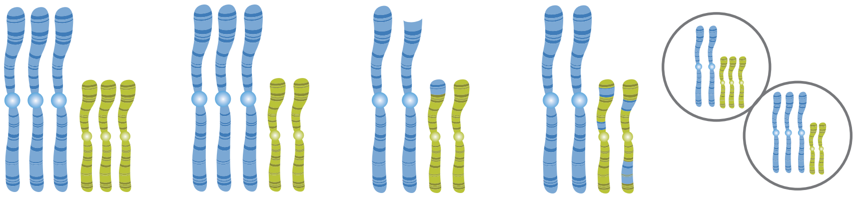
Highly Reproducible Data

All Infinium products have high feature redundancy and contain sample-dependent and sample-independent internal controls, so you can have complete confidence in your data. Multi-sample formats supported by high-density arrays minimize array-to-array processing variability, reduce handling errors, and allow more efficient scanning.

Quality-controlled Arrays

Unlike oligo arrays that are synthesized one at a time, Infinium bead pools are shared across months' worth of product, providing consistency from one lot to the next. During the manufacturing process, SNPs on every BeadChip are positionally verified. All arrays must pass a robust quality control before shipping.

DETECT A WIDE RANGE OF CHROMOSOMAL ABNORMALITIES



	MEDIAN MARKER SPACING	POLYPLOIDY	ANEUPLOIDY	UNBALANCED ABERRATION	AMPLIFICATION/ DUPLICATION	LOW-LEVEL MOSAICISM	COPY-NEUTRAL LOH
Infinium BeadChips	0.64 kb*–6.2 kb**	✓	✓	✓	✓	✓	✓

* Infinium HumanOmni2.5-8 BeadChip
** Infinium HumanCytoSNP-12 BeadChip

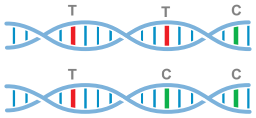
Robust assay. Simplified workflow.

Ease of use. Automation options. The Infinium Assay features single-tube sample prep and amplification without PCR or ligation steps. Our streamlined protocol and reagent sets save you time and reduce sample handling errors. And since we use familiar molecular biology techniques, transitioning from your existing technology is easy.

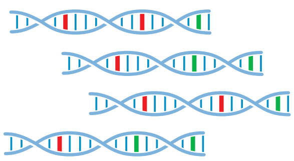
The three-day assay protocol requires less than fours hours hands-on time to process four multi-sample BeadChips (less than one hour for automated processing). For high-throughput operation, we offer modular automation options. A customized liquid-handling robot provides assay automation, and the AutoLoader 2.x allows 24-hour-per-day unattended scanning. Add Illumina LIMS control to maximize accuracy and throughput.

SAVE TIME WITH AN EASY-TO-USE WORKFLOW

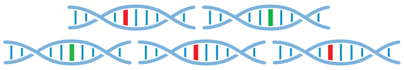
1 GENOMIC DNA (200–400 ng)



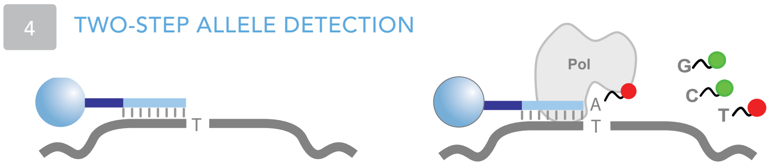
2 LINEAR WHOLE-GENOME AMPLIFICATION



3 FRAGMENT DNA



4 TWO-STEP ALLELE DETECTION



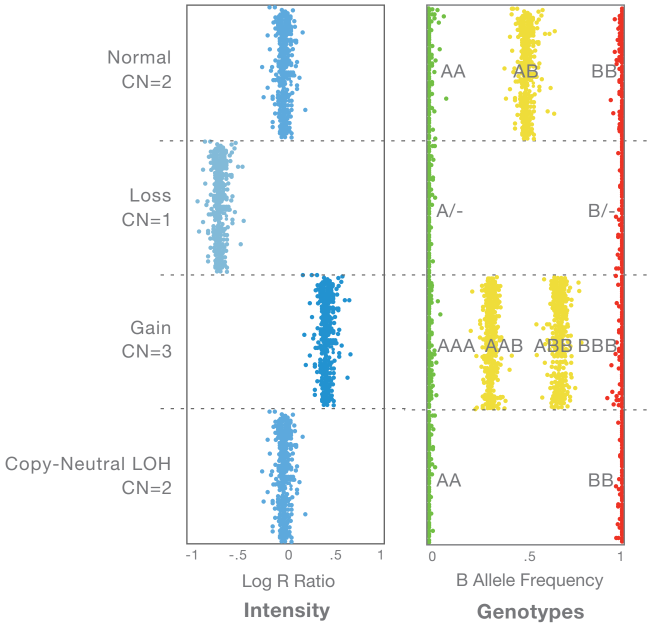
Step 1. Selectivity

Hybridization of unlabeled DNA fragment to 50mer probe on array

Step 2. Specificity

Enzymatic single-base extension with labeled nucleotide

5 COPY NUMBER ANALYSIS



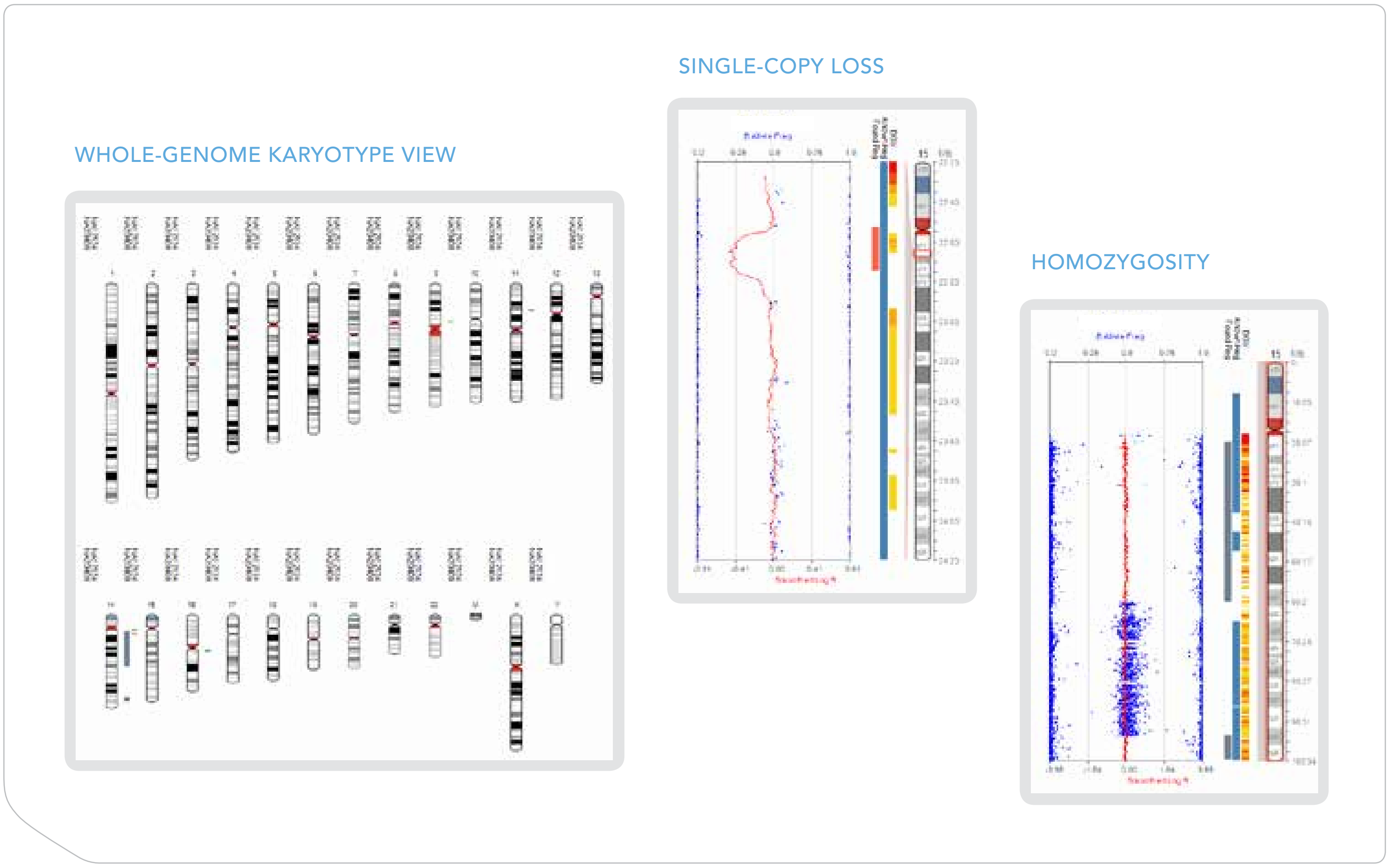
Efficient identification. Intuitive analysis.

Customized views. Convenient software. Developed in collaboration with molecular cytogeneticists, our KaryoStudio software package offers user-friendly, intuitive analysis of Infinium data.

Quickly and efficiently identify important structural aberrations, match these to known aberrations, and generate reports based on your findings. Customize analysis by using a standard region list or specific regions and phenotypes for cross-matching. Link out to external resources, such as the Database of Genomic Variants, UCSC genome browser, and DECIPHER. KaryoStudio gives you the right set of features in a streamlined workflow.

For more information about KaryoStudio, including a demonstration video, visit www.illumina.com/karyostudio

RAPIDLY SCAN FOUND REGIONS WITH KARYOSTUDIO



WHOLE-GENOME KARYOTYPE VIEW

SINGLE-COPY LOSS

HOMOZYGOSITY

Worldwide coverage. Personalized service.

We partner with you to complete the installation and training process, and this personalized support continues long after your initial training. One call to our Technical Support group puts you in touch with a team of Ph.D. scientists who are experts on Illumina systems, assays, and software. Our dedicated service and support teams give you peace of mind to focus on the biology.

To learn more, visit www.illumina.com/support

www.illumina.com

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