Genome-Wide DNA Analysis BeadChips

Illumina has created a comprehensive portfolio of DNA Analysis tools by deploying industryleading content on multi-sample Infinium[®] HD BeadChips. Unmatched power provides researchers the fastest path to discoveries and publication.

COMPREHENSIVE ANALYSIS PLATFORM

Illumina is a leader in the field of genetic analysis with innovative tools for DNA analysis, RNA analysis, and high-throughput sequencing. With the newest generation of high-density Infinium HD products, Illumina continues to provide the most comprehensive and powerful family of DNA Analysis BeadChips, taking genotyping and copy number variation (CNV) analysis to the next level.

Infinium HD technology expands the limits of density to provide industry-leading multiplexing in multi-sample formats, while maintaining the high data quality and

INFINIUM HD BEADCHIP HIGHLIGHTS

- Proven Content: Publish with confidence using a foundation of well-validated assays and markers
- Powerful Cytogenetics: Get high-resolution analysis with dense and uniform marker spacing with minimal gaps
- High Density: Assay nearly 1.2 million loci per sample
- Multi-Sample Format: Increase sample throughput to finish projects faster
- Low Sample Input: Interrogate limited sample sources, down to 200 ng of DNA per sample

simple assay workflow common to all Illumina products. Furthermore, Infinium HD BeadChips have low DNA input requirements, expanding the range of sample sources that can be used for a study.

The Infinium HD products include the HumanCytoSNP-12, Human660W-Quad, Human1M-Duo, and HumanOmni1-Quad BeadChips (Figure 1). This family of Illumina BeadChips provides a broad spectrum of whole-genome DNA Analysis products to support a variety of experimental designs. Researchers have the flexibility to use panels of 300,000 to nearly 1,200,000 markers per sample, depending on their study goals. All of these BeadChips provide powerful and integrated genomewide SNP genotyping and structural variant detection. The 12-sample HumanCytoSNP-12 is a streamlined whole-genome scanning panel for high sample throughput analysis of genetic and structural variation, including cytogenetic abnormalities. The powerful Human660W-Quad BeadChip has an ideal combination of high-coverage genome-wide SNP and CNV markers in a highthroughput format. The two-sample Human1M-Duo BeadChip provides comprehensive access to the genome with nearly 1.2 million markers covering genome-wide SNPs,



Infinium HD BeadChips provide a broad range of powerful content options in high-throughput formats for processing two, four, or 12 samples simultaneously.

CNV-targeted markers, and high-value functional regions. The four-sample HumanOmni1-Quad offers the best combination of power and throughput, featuring over one million strategically selected markers that deliver dense genome-wide coverage and extensive disease-associated content, including data from the 1000 Genomes Project. The unparalleled content and assay technology of Infinium HD BeadChips provide the fastest path to discoveries and publication.

POWERFUL MARKERS

Genome-wide association studies (GWAS) rely on genotyping SNPs near a disease locus to identify genetic links to disease. As highlighted in a study from University of Michigan re-



searchers comparing different array platforms, Illumina's marker selection strategy is demonstrably better for GWAS studies¹. Infinium Bead-Chips offer benefits in terms of several critical parameters that together contribute to the statistical power in an experiment: genomic coverage, array efficiency, genic coverage, call rate, and call accuracy².

The power to detect an association depends on the linkage disequilibrium (r²) between the genotyped marker and the adjacent diseasecausing SNP. A high r² between two SNPs indicates that the two SNPs can act as good proxies (tag SNPs) for each other³. Because the Infinium HD Assay chemistry—like the Infinium II Assay—affords flexible marker selection, Illumina scientists are able to rationally select loci that provide the highest information content, while using fewer SNPs. Illumina has taken advantage of this flexibility by selecting powerful tag SNPs and other high-value regions for markers. A result of this strategy is that the ~300,000 markers on the HumanCytoSNP-12 provide nearly the same genomic coverage in the Caucasian (CEU) population as a competing 924,000-marker array.

Compared to microarrays with randomly selected SNP content, Illumina's DNA Analysis BeadChips offer the industry's highest statistical power per sample by reducing the correction factor for multiple testing by almost 40%. Higher power means fewer samples are needed to identify significant genetic variations. Studies can be completed faster and more economically to support rapid publication in top-tier journals (for examples, browse customer citations at www.illumina.com/publications).

COMPREHENSIVE COVERAGE

Illumina DNA Analysis BeadChips provide optimized panels for surveying genetic variants^{1,4}. All genome-wide Infinium DNA Analysis products start with a broad set of tag SNPs and other valuable SNPs from the International HapMap Project and NCBI's dbSNP to provide high genomic coverage and uniformity across the genome. All genome-wide DNA Analysis products also include a set of additional CNV-targeted markers designed to increase coverage of regions underrepresented by tag SNPs.

In the Illumina portfolio, individual BeadChips offer slightly different content and numbers of markers to provide flexible options for using the optimal content panel in any study design (Table 1).

HumanCytoSNP-12 DNA Analysis BeadChip Content

The HumanCytoSNP-12 BeadChip represents the most efficiency-optimized DNA Analysis content selection strategy. It includes a complete panel of genome-wide tag SNPs and additional markers targeting all regions of known cytogenetic importance.

Illumina scientists employed 200,000 "best of the best" SNPs that have the highest tagging power. This content maintains the exceptional genome-wide SNP coverage that Illumina is known for (70% in CEU at $r^2 > 0.8$) because of the efficient marker design strategy². At the same time, a set of 220,000 markers provides extra utility for cytogenetic analysis. This includes dense coverage of ~250 genomic regions commonly studied in cytogenetics labs and targeted coverage in ~400 additional genes, subtelomeric regions, pericentromeric regions, and sex chromosomes⁵.

Furthermore, the HumanCytoSNP-12 takes advantage of the industry's first 12-sample whole-genome BeadChip and Illumina's high-density array technology to provide the highest throughput and most cost-effective BeadChip.

Human660W-Quad DNA Analysis BeadChip Content

The Human660W-Quad BeadChip offers comprehensive genomic coverage across many populations and the majority of known variation



The Human660W-Quad BeadChip content covers the majority of common variation in three distinct populations. Graphs are estimated, based on the HapMap release 24 data set of > 2.3 million common SNPs.

in regions of the genome based on HapMap data.

The Human660W-Quad BeadChip builds on the content of the highly successful HumanHap550 BeadChip. The broad, evenly spaced wholegenome marker set provides high genomic coverage for powerful GWAS. In addition, the Human660W-Quad BeadChip provides 87%, 85%, and 56% coverage of CEU, CHB+JPT, and YRI populations at r² > 0.8 (figure 2).

For equally powerful CNV and cytogenetic analysis, this dense backbone content is combined with an additional ~100,000 markers that target observed common CNVs.

The entire panel of 657,000 markers provides exceptional genomic coverage and identification of known and novel structural variants, combined with an efficient multi-sample format.

Human1M-Duo DNA Analysis BeadChip Content

With nearly 1.2 million markers per sample, the Human1M-Duo provides

a powerful combination of quality, coverage, and throughput. The comprehensive set of markers on the Human1M-Duo BeadChip provides access to dense genome-wide tag SNP coverage as well as additional content targeted to high-value genomic regions of interest. Other probes are located in SNP deserts to fill in gaps.

The uniform genome-wide coverage results in a median spacing between markers of 1.5 kb (mean = 2.4 kb) and few large gaps for high-resolution CNV identification and cytogenetics analysis. Ensuring no regions are skipped, the 90th percentile largest gap between SNPs on the Human1M-Duo BeadChip is 6 kb. The result of this comprehensive design strategy is 95%, 93%, and 76% coverage of CEU, CHB+JPT, and YRI populations at r² > 0.8.

In addition to the broad coverage crucial for successful whole-genome association studies, the Human1M-Duo BeadChip targets other highvalue content. Gene-centric markers



The HumanOmni1-Quad BeadChip content covers the majority of HapMap common variation in three distinct populations. Graphs are based on the HapMap release 26 data set of > 2.3 million common SNPs.

selected in and around genes target both synonymous and non-synonymous SNPs to increase genic coverage. In addition, more than 10,000 markers are included for the major histocompatibility complex (MHC) region, which contains a high density of genes often associated with autoimmune and infectious diseases.

The BeadChip also features ~60,000 CNV-targeted markers, developed in collaboration with deCODE Genetics, for regions likely to contain undiscovered CNV. Novel CNV-specific probes and the dense uniform genome-wide SNP coverage support unbiased discovery and analysis of copy number polymorphisms.

HumanOmni1-Quad DNA Analysis BeadChip Content

The HumanOmni1-Quad BeadChip provides an unparalleled, extensive view of the genome, in a highthroughput, cost-effective format. A complete optimization of the BeadChip design increases the available complexity, allowing nearly five million markers to be assayed across four different samples in parallel, while reducing the amount of required DNA to 200 ng.

Each BeadChip features over one million available assays per sample, containing carefully selected content that delivers dense coverage of the human genome and targets regions known to play a role in human disease. This comprehensive collection of genomic markers offers the best combination of power, price and throughput available for genomewide association studies.

With recently released data from all three HapMap phases, intelligent tag SNP selection has been optimized to maintain comprehensive genomic coverage, while reducing SNP redun-

TABLE 1: COMPREHENSIVE COVERAGE OF HIGH-VALUE REGIONS

	HUMANCYTOSNP-12 v2	HUMAN660W-QUAD v1	HUMAN1M-DUO v3	HUMANOMNI1-QUAD v1
Overview	Efficient coverage for cost-effective GWAS and cytogenetic screening	High genomic coverage of common SNPs and CNV regions	Genome-wide cover- age and additional high-value regions	Comprehensive genome- wide coverage and additional high-value regions, including new content from 1,000 Genomes Project
Number of Markers per Sample	301, 232	657,366	1,199,187	1,140,419
Number of Samples per BeadChip	12	4	2	4
DNA Input Requirement (per sample)	200 ng	200 ng	400 ng	200 ng
Scan Times per Sample (minutes) [#]	3	9	18	13
Genomic Coverage				
CEU (Mean / Median / r² > 0.8)	0.81 / 0.94 / 0.70	0.92 / 1.0 / 0.87	0.96 / 1.0 / 0.95	0.95 /1.0 / 0.93
CHB+JPT	0.83 / 0.94 / 0.73	0.92 / 1.0 / 0.85	0.95 / 1.0 / 0.93	0.94 / 1.0 / 0.92
YRI	0.55 / 0.52 / 0.32	0.74 / 0.87 / 0.56	0.86 / 1.0 / 0.76	0.85 / 1.0 / 0.76
Minor Allele Frequency*				
CEU (Mean / Median)	0.22 / 0.21	0.24 / 0.23	0.20 / 0.18	0.19 / 0.17
CHB+JPT	0.21 / 0.20	0.21 / 0.20	0.18 / 0.16	0.18 / 0.15
YRI	0.21 / 0.19	0.22 / 0.21	0.20 / 0.17	0.20 / 0.18
Spacing (kb)				
(Mean / Median)	9.6 / 6.2	4.4 / 2.3	2.4 / 1.5	2.4/ 1.2
90th %ile Largest Gap	18.6	10.6	6.0	6.4
Marker Categories				
Markers Within 10 kb of a RefSeq Gene	148,666	332,756	672,002	618,959
Non-Synonymous SNPs§	3,480	10,051	21,877	32,110
MHC [†] / ADME [‡] / Indel SNPs	761 / 2,382 / 0	3,177 / 8,440 / 0	10,415 / 20,493 / 483	19,081 / 22,429 / 459
Sex Chromosome (X / Y / PAR Loci)	15,063 / 2,841 / 1,579	16,509 / 44 / 15	45,591 / 4,637 / 979	27,493 / 2,322 / 1,157
Mitochondrial SNPs	0	135	138	27

Scan times are approximations based on the iScan platform

* Based on HapMap rel 24 for HumanCytoSNP-12, Human660W-Quad, and HumanOmni1-Quad, and rel 23 for Human1M-Duo § Based on RefSeq and Ensembl databases

[†] As defined by de Bakker, 2006
[‡] Within 10 kb of 333 known ADME-related gene



in substantially better coverage of important regions compared to greater numbers of randomly selected markers.

dancy. This has enabled the inclusion of additional content carefully chosen to target high-value regions of the genome, such as the MHC region and new coding variants identified by the 1000 Genomes Project. The redesigned SNP selection strategy has maintained high genomic coverage rates of 93%, 92%, and 76% at $r^2 > 0.8$ for the CEU, CHB+JPT, and YRI populations, respectively (Figure 3). High density markers with a median spacing of 1.5 kb and the fewest number of large gaps for any BeadChip ensure the highest level of resolution for CNV identification in the industry.

The HumanOmni1-Quad is the only BeadChip to include cuttingedge content derived from the 1000 Genomes Project. This large international effort is dramatically increasing the information we have about genetic variation across human populations⁶. Already, the project has uncovered millions of rare and novel SNPs that will drive the next generation of microarrays. For the HumanOmni1-Quad, SNPs selected from the 1000 Genomes Project focus on regions already identified in GWAS to be associated with human disease. This content includes ~18,000 SNPs targeting four 1Mb regions known to be associated to three or more human diseases; over 50,000 SNPs predicted to be nonsynonymous; 62,000 SNPs covering an additional 100 intervals surrounding published peak markers from the NHGRI GWAS database; and the remaining 950 top single-marker associated SNPs from the GWAS database.

With high-throughput processing, comprehensive genomic coverage and the ability to capture a vast amount of genetic variation, the HumanOmni1-Quad BeadChip lets you make more meaningful discoveries and take the fastest path to publication.

SENSITIVE STRUCTURAL VARIANT DETECTION

Dense Uniform Markers

An important goal during the design of Infinium HD content panels was the uniform distribution of SNP markers to create the best panels for detecting structural variation, including loss of heterozygosity. With the fewest large gaps across the whole genome, the HumanOmni1-Quad BeadChip is an ideal tool for CNV researchers to use for discovery and high-resolution breakpoint mapping (Figure 4).

Intelligent Targeted Content

Of course, some regions of the genome are naturally underrepresented by tag SNPs. Illumina scientists have leveraged the flexible Infinium Assay design to generate marker sets that provide the industry's best CNV detection panels.

The HumanCytoSNP-12 BeadChip is optimized to efficiently detect cytogenetic abnormalities that are the most relevant to human disease. Its content panel targets common regions shown to be important for cytogenetic analysis⁵ and a dense backbone of coverage across the remainder of the genome.

The Human660W-Quad contains a set of ~100,000 markers that are highly informative for analyzing common CNV regions. These markers were identified in a high-density screen for CNVs that occur in two or more HapMap samples, which was conducted in collaboration with The Centre for Applied Genomics at the Hospital for Sick Children in Toronto, the Wellcome Trust Sanger Institute in the United Kingdom, and Harvard Medical School/Brigham and Women's Hospital in Boston.

The Human1M-Duo features content developed in collaboration with deCODE Genetics to blanket the "unSNPable genome" with additional non-polymorphic markers⁷. This includes difficult-to-analyze regions like megasatellites and segmental duplications, which are targeted with both SNPs and non-polymorphic probes. Many of these regions have been validated with other approaches, such as TaqMan and Southern blotting, to confirm variance in copy number in several representative populations.

The HumanOmni1-Quad includes extensive high-value content focused on disease-associated regions: cSNPs, eSNPs, indels, SNPs in mRNA splice sites, miRNA binding sites, introns, promoter regions, ADME genes, disease-associated SNPs, mitochondrial DNA, AIMs, ABO blood typing SNPs, PAR, Y-chromosome, MHC region, and HLA complex. The BeadChip also provides high CNV coverage (*Figure 4*), featuring 5,000+ rare CNV regions in addition to all the common CNV content available on the Human660W-Quad.

CNV-targeted probes share the same rational design strategy with all SNPs. All markers on Infinium HD BeadChips have high feature redundancy, yielding low overall noise, and all markers are used for reliable and sensitive detection of changes in copy number. The consistent marker design allows all markers to be analyzed together using GenomeStudio® Software. Completely integrated genotyping and copy number studies maximize analytical efficiency⁸⁻¹⁰.

The resulting rationally designed content on Infinium HD BeadChips supports the industry's most powerful SNP genotyping and CNV identification^{3,1}.

CUSTOM CONTENT OPTIONS

Illumina offers the option of adding custom-designed content to the broad genome-wide standard SNP content on the Human1M-Duo and Human660W-Quad BeadChips. The results are semi-custom Human1M-Duo+ and HumanHap550-Quad+ BeadChips. With assistance from Illumina scientists and a proprietary Assay Design Tool, researchers can include an additional panel of up to 60,800 SNPs to the powerful standard content.

STREAMLINED ASSAY WORKFLOW

The Infinium HD Assay can be scaled to unlimited multiplexing without compromising data quality, unlike many alternative PCR-dependent assays. The simple, streamlined workflow is common across all products, no matter how many SNPs are being interrogated. Likewise, the data acquisition process and analysis are the same.

The Infinium HD Assay protocol (Figure 5) features single-tube sample preparation and whole-genome amplification without PCR or ligation steps, significantly reducing labor and sample handling errors. After hybridizing unlabeled DNA sample to the BeadChip, two-step allele detection provides high call rates and accuracy. Selectivity and specificity are accomplished in two steps. Target hybridization to bead-bound 50-mer oligos provides high selectivity while enzymatic single-base extension provides powerful specificity. The singlebase extension also incorporates a labeled nucleotide for assay readout. The staining reagent is optimized to provide a higher signal, and more balanced intensities between red and green channels. These features contribute to industry-leading accuracy, high call rates, and copy number data with lower noise.

The iScan System uses advanced optics for high-resolution detection and high-throughput readout of assay results. With this system and 12-sample BeadChips, researchers can scan each sample in three minutes (Table 1).



Multi-Sample Format

The efficient multi-sample format of Illumina BeadChips cost-effectively increases sample throughput. Reduced handling, more efficient scanning, and higher density assays contribute to higher sample throughput rates so projects are finished faster. Also, by effectively eliminating array-to-array variability, the multisample format is ideal for analyzing matched samples.

ILLUMINA® DNA ANALYSIS

Low DNA Input Requirement

Infinium HD BeadChips require low quantities of input DNA, providing opportunities to use more limited sample sources (*Table* 1). Four- or 12-sample Infinium HD BeadChips require only 200 ng DNA per sample, and two-sample BeadChips require 400 ng DNA per sample.

HIGH QUALITY DATA

All of the assays on the HumanOmni1-Quad, Human1M-Duo, Human660W-Quad, and HumanCytoSNP-12 DNA Analysis BeadChips use Infinium HD chemistry. These BeadChips have undergone the same rigorous functional testing that ensures strong and reproducible performance of all Illumina products. One assessment of data quality was the analysis of a diverse panel of HapMap reference samples (*Table 2*). As shown in Table 2, the Infinium HD BeadChips perform extremely well, producing high call frequencies and excellent reproducibility.

Successful genome-wide associa-

TABLE 2: GENOTYPING DATA QUALITY OF DNA ANALYSIS BEADCHIPS USING REFERENCE SAMPLES

Genotyping Parameter	Value from Reference Samples	Product Specification	CNV Analysis Parameter	Value from Reference Samples	Expected**			
Call Frequency	99.71%	> 99% average	$Log R Ratio^{\dagger}$	0.14	< 0.30			
Reproducibility	100.00%	> 99.9%	B Allele Frequency ^{†§}	0.03	< 0.04			
Mendelian Inconsistencies	0.02%	< 0.1%						
HapMap Concordance	99.25%	N/A						
HUMAN660W-QUAD BEADCHIP (283 DNA SAMPLES, 15 REPLICATES, 58 TRIOS)								
Genotyping Parameter	Value from Reference Samples	Product Specification	CNV Analysis Parameter	Value from Reference Samples	Expected**			
Call Frequency	99.96%	> 99% average	Log R Ratio [†]	0.16	< 0.30			
Reproducibility	100.00%	> 99.9%	B Allele Frequency ^{†§}	0.03	< 0.04			
Mendelian Inconsistencies	0.04%	< 0.1%						
HapMap Concordance	99.76%	N/A						
HUMAN1M-DUO BEADCHIP (284 DNA SAMPLES, 15 REPLICATES, 58 TRIOS)								
Genotyping Parameter	Value from	Product	CNV Analysis Parameter	Value from	Exported**			
	Reference Samples	Specification	5	Reference Samples	Expected			
Call Frequency	Reference Samples 99.83%	Specification > 99% average	Log R Ratio [†]	0.15	< 0.30			
Call Frequency Reproducibility	Reference Samples 99.83% 100.00%	Specification > 99% average > 99.9%	Log R Ratio [†] B Allele Frequency ^{†§}	0.15 0.03	< 0.30 < 0.04			
Call Frequency Reproducibility Mendelian Inconsistencies	Reference Samples 99.83% 100.00% 0.05%	Specification > 99% average > 99.9% < 0.1%	Log R Ratio [†] B Allele Frequency ^{†§}	0.15 0.03	< 0.30 < 0.04			
Call Frequency Reproducibility Mendelian Inconsistencies HapMap Concordance	Reference Samples 99.83% 100.00% 0.05% 99.63%	Specification > 99% average > 99.9% < 0.1% N/A	Log R Ratio [†] B Allele Frequency ^{†§}	0.15 0.03	< 0.30 < 0.04			
Call Frequency Reproducibility Mendelian Inconsistencies HapMap Concordance	Reference Samples 99.83% 100.00% 0.05% 99.63%	Specification > 99% average > 99.9% < 0.1% N/A MPLES, 15 REPLIC	Log R Ratio [†] B Allele Frequency ^{†§}	0.15 0.03	< 0.30 < 0.04			
Call Frequency Reproducibility Mendelian Inconsistencies HapMap Concordance HUMANOMNI1-QUAD BE Genotyping Parameter	Reference Samples 99.83% 100.00% 0.05% 99.63% ADCHIP (282 DNA SA Value from Reference Samples	Specification > 99% average > 99.9% < 0.1% N/A MPLES, 15 REPLIC Product Specification	Log R Ratio [†] B Allele Frequency ^{†§} CATES, 56 TRIOS) CNV Analysis Parameter	Value from Reference Samples	< 0.30 < 0.04 Expected**			
Call Frequency Reproducibility Mendelian Inconsistencies HapMap Concordance HUMANOMNI1-QUAD BEA Genotyping Parameter Call Frequency	Reference Samples 99.83% 100.00% 0.05% 99.63% ADCHIP (282 DNA SA Value from Reference Samples 99.87%	Specification > 99% average > 99.9% < 0.1% N/A MPLES, 15 REPLIC Product Specification > 99% average	Log R Ratio [†] B Allele Frequency ^{†§} CATES, 56 TRIOS) CNV Analysis Parameter Log R Ratio [†]	Value from Reference Samples 0.15 0.03	 < 0.30 < 0.04 Expected** < 0.30 			
Call Frequency Reproducibility Mendelian Inconsistencies HapMap Concordance HUMANOMNI1-QUAD BE Genotyping Parameter Call Frequency Reproducibility	Value from Reference Samples 99.83% 100.00% 0.05% 99.63% ADCHIP (282 DNA SA Value from Reference Samples 99.87% 100.00%	Specification > 99% average > 99.9% < 0.1% N/A MPLES, 15 REPLIC Product Specification > 99% average > 99.9%	Log R Ratio [†] B Allele Frequency ^{†§} CATES, 56 TRIOS) CNV Analysis Parameter Log R Ratio [†] B Allele Frequency ^{†§}	Value from Reference Samples	 < 0.30 < 0.04 Expected** < 0.30 < 0.04 			
Call Frequency Reproducibility Mendelian Inconsistencies HapMap Concordance HUMANOMNI1-QUAD BE Genotyping Parameter Call Frequency Reproducibility Mendelian Inconsistencies	Reference Samples 99.83% 100.00% 0.05% 99.63% ADCHIP (282 DNA SA Value from Reference Samples 99.87% 100.00% 0.02%	Specification > 99% average > 99.9% < 0.1%	Log R Ratio [†] B Allele Frequency ^{†§} CATES, 56 TRIOS) CNV Analysis Parameter Log R Ratio [†] B Allele Frequency ^{†§}	Value from Reference Samples	 < 0.30 < 0.04 Expected** < 0.30 < 0.04 			

* Based on CEU trios using loci with MAF \geq 0.01; given as the frequency of markers with minor allele undertransmitted relative to the expected 50%

** Values expected for typical projects, excluding tumor samples or any samples prepared not following standard Illumina protocols

[†] Excludes sex chromosomes, mtDNA, and intensity-only loci

§ Heterozygotes only

tion studies depend, in part, on the high call rates that Illumina DNA Analysis BeadChips exhibit. Since complex disease traits often have relatively small gene effects, potential associations may be missed if an assayed SNP, in LD with a disease SNP, has a low call rate. Data from the Infinium HD DNA Analysis BeadChips show strong reproducibility (> 99.9%) and concordance with the International HapMap Project (> 99.2%). Additionally, these BeadChips provide precise copy number metrics with low overall noise levels (Table 2), allowing reliable detection of single changes in copy number.

Internal Quality Controls

All products based on the Infinium HD Assay have several sample-dependent and sample-independent internal controls so researchers have confidence that they are producing the highest quality data. The performance of all controls can be monitored easily with the GenomeStudio Genotyping Module integrated Controls Dashboard.

ANALYSIS SOFTWARE

Illumina's GenomeStudio Data Analysis Software offers integrated genotyping and copy number tools and a graphical Genome Viewer. GenomeStudio Software has an open plug-in interface to integrate thirdparty applications for more downstream data analysis options. The illumina•Connect¹¹ program leverages this open architecture and has made numerous plug-ins available to support genotyping and copy number analysis.

iControlDB

Illumina hosts a database of genotypic and phenotypic data generated by researchers using Illumina genotyping products, which can be used to supplement controls in case-control association studies¹². Access to the thousands of controls in the free iControlDB database allow researchers to increase the power of an association study and decrease overall project costs.

AUTOMATION

As with most of Illumina's standard DNA Analysis products, an optional Laboratory Information Management System (LIMS) and robotic automation accurately and efficiently track samples to provide workflow management and overall project management. This system, custom designed for Infinium workflows, allows labs to maximize their throughput with a completely integrated microarray solution.

SERVICES

Illumina FastTrack Genotyping Services are available to analyze samples in a timely fashion at a reasonable cost using any Infinium DNA Analysis BeadChip. This option allows researchers to acquire high-quality data for limited studies or before purchasing their own equipment.

SUMMARY

Illumina whole-genome DNA Analysis BeadChips are high-quality tools for SNP genotyping and analysis of structural variants. This genetic analysis platform offers a range of solutions with different numbers of markers per sample and different numbers of samples per BeadChip. All Illumina Bead-Chips offer the highest data quality and most complete genomic coverage in the industry. By choosing a BeadChip that matches the study design, researchers can confidently pursue the fastest path to discoveries and publication.

ILLUMINA® DNA ANALYSIS

ORDERING INFORMATION

PRODUCTBEADCHIPSSAMPLESCATALOG NO.HumanCytoSNP-12 v2 BeadChips and Reagents112WG-320-0202448WG-320-020324288WG-320-0204961,152WG-320-0205416WG-311-1501Human660W-Quad v1 BeadChips and Reagents1248WG-311-150296384WG-311-150396384WG-311-1503Human1M-Duo v3 BeadChips and Reagents2448WG-311-1102192384WG-311-1103192384WG-311-1104Human0mni1-Quad v1 BeadChips and Reagents416WG-311-111011248WG-311-111011248WG-311-11101Human0mni1-Quad v1 BeadChips and Reagents2496WG-311-111011248WG-311-111011248WG-311-111011248WG-311-11111248WG-311-11111248WG-311-11111248WG-311-11111248WG-311-11111248WG-311-1111131416WG-311-11111248WG-311-11111496384WG-311-11111248WG-311-1111			_	
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BeadChips and Reagents 24 96 WG-311-1112 96 384 WG-311-1113	HumanOmni1-Quad v1	12	48	WG-311-1111
96 384 WG-311-1113	BeadChips and Reagents	24	96	WG-311-1112
		96	384	WG-311-1113

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