Infinium™ CoreExome-24 v1.3 BeadChip

Customizable, high-density array for cost-effective, large-scale genotyping and screening studies.

Overview

The customizable Infinium CoreExome-24 v1.3 BeadChip offers an economical way to perform and support large genetic studies, especially large-scale genotyping studies. Developed in collaboration with several leading research institutions, the Infinium CoreExome-24 v1.3 BeadChip includes all the tag single nucleotide polymorphisms (SNPs) found on the Infinium Core-24 BeadChip, plus over 240,000 markers from the Infinium HumanExome BeadChip (Table 5 and Table 6). The Infinium CoreExome-24+ v1.3 BeadChip has the added capacity to include up to 100,000 semicustom markers. In addition to performing cost-effective large-scale genotyping studies, the Infinium CoreExome-24 v1.3 BeadChip can be used to obtain baseline sample data sets for various downstream applications quickly and easily. These applications include common variant, mitochondrial DNA (mtDNA), ancestry, sex confirmation, loss of-variant, and insertion/deletion (indel) detection studies. Infinium CoreExome-24 v1.3 BeadChips use the trusted Infinium high-throughput screening (HTS) Assay. When combined with the proven iScan™ or HiScan™ System, this high-density, 24-sample BeadChip (Figure 1) delivers affordable, high-quality, genome-wide information across diverse world populations.

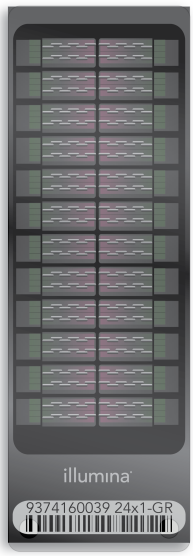

Figure 1: The Infinium CoreExome-24 v1.3 BeadChip— The Infinium CoreExome-24 v1.3 BeadChip enables informative genotyping of tag SNP and exome-focused markers across diverse world populations, delivering high-quality data that can be used for various downstream applications.

High-throughput workflow

The Infinium CoreExome-24 v1.3 BeadChip uses the highly scalable 24-sample Infinium HTS format for high-throughput processing of thousands of samples per week for large, population-scale research and variant screening. The Infinium HTS format also provides a rapid three-day workflow that allows genotyping service providers and clinical researchers to gather data and advance studies quickly (Figure 2).

Optional integration of the Illumina Laboratory Information Management System (LIMS) into the workflow provides high laboratory efficiency with automation functionality, process tracking, and quality control (QC) data tracking. The Illumina ArrayLab Consulting Service offers customized solutions to high-throughput genotyping labs that desire increased efficiency and overall operational excellence.

Robust, high-quality assay

The Infinium CoreExome-24 v1.3 BeadChip uses proven Infinium assay chemistry to deliver the same high-quality, reproducible data (Table 1) that Illumina genotyping arrays have provided for over a decade. The Infinium product line provides high call rates and high reproducibility for numerous sample types including, saliva, blood, solid tumors, fresh frozen, and buccal swabs (Table 2–Table 4). In addition, the high signal-to-noise ratio of the individual genotyping calls from the Infinium assay provides researchers with access to genome-wide copy number variant (CNV) calling with a mean probe spacing of ~ 5.27 kb.

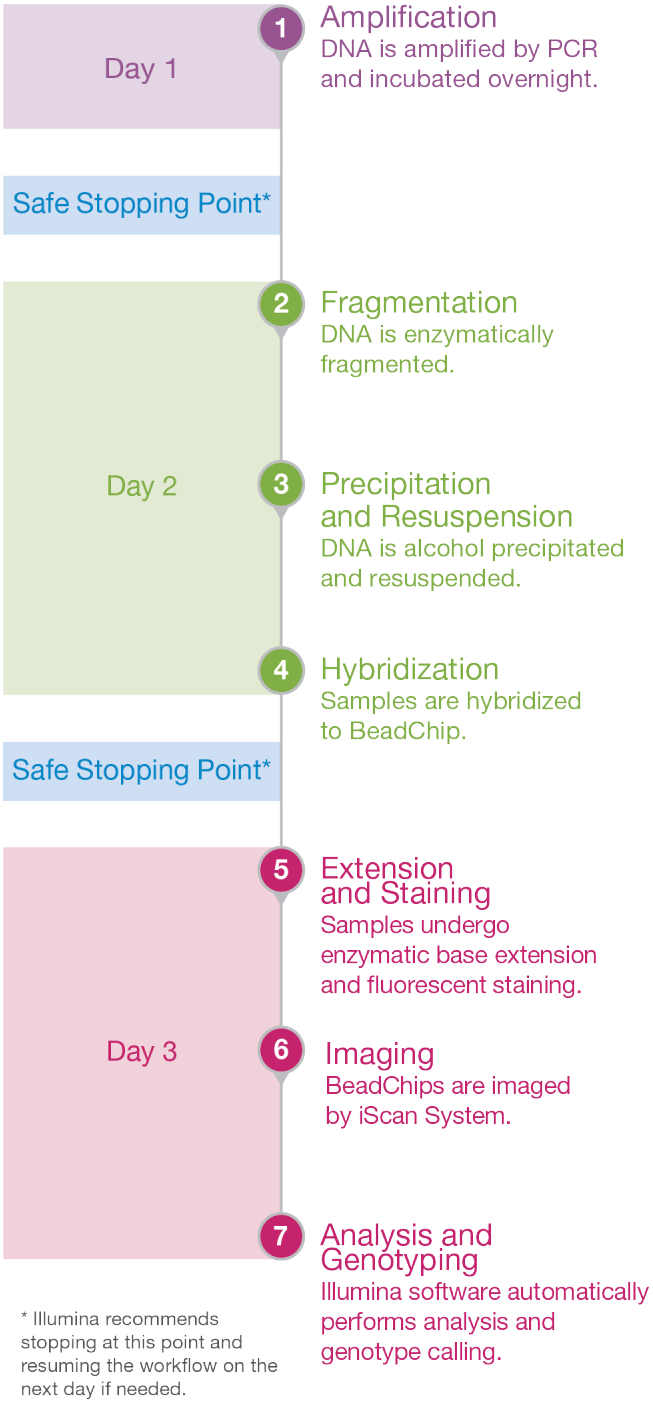

Figure 2: The Infinium HTS workflow–The Infinium HTS format provides rapid 3-day workflow with minimal hands-on time.

Table 1: Product information

Feature	Description		
Species	Human		
Total number of markers	551,004		
Capacity for custom bead types	100,000		
Number of samples per BeadChip	24 Samples		
DNA input requirement	200 ng		
Assay chemistry	Infinium HTS		
Instrument support	iScan or HiScan System		
Sample throughput[a]	~ 2304 samples/week		
Scan time per sample	iScan System 2.5 min	HiScan System 2.0 min	
Data performance	**Value[b]**	**Product Specification[d]**	
Call rate	99.8%	> 99% avg.	
Reproducibility	99.99%	> 99.9%	
Log R deviation	0.09	< 0.30[c]	
Spacing			
Spacing (kb)	Mean 5.27	Median 1.82	90th%[c] 14.30

a. Estimate assumes 1 iScan System, 1 AutoLoader 2.x, 2 Tecan robots, and a 5-day work week.
b. Values are derived from genotyping 333 HapMap reference samples.
c. Value expected for typical projects using standard Illumina protocols. Tumor samples and samples prepared by methods other than standard Illumina protocols are excluded.
d. Excludes Y chromosome markers for female samples.

Table 2: Imputation accuracy from 1000G[a] at various MAF thresholds

Population[b]	Imputation accuracy		
	MAF ≥ 5%	MAF ≥ 1%	MAF 1–5%
AFR	0.90	0.84	0.76
AMR	0.94	0.89	0.80
EAS	0.93	0.86	0.66
EUR	0.94	0.89	0.76
SAS	0.93	0.86	0.71

a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). www.1000genomes.org. Accessed July 2016.
b. See www.1000genomes.org/category/frequently-asked-questions/population

Abbreviations: MAF: minor allele frequency; AFR: African; AMR: Ad Mixed American; EAS: East Asian; EUR: European; SAS: South Asian.

Table 3: LD r^2 ≥ 0.80 from 1000G[a] at various MAF thresholds

1000G population[b]	LD coverage (r^2 ≥ 0.80)	
	MAF ≥ 5%	MAF ≥ 1%
AFR	0.29	0.18
AMR	0.57	0.40
EAS	0.66	0.54
EUR	0.63	0.49
SAS	0.58	0.44

a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). www.1000genomes.org. Accessed July 2016.
b. See www.1000genomes.org/category/frequently-asked-questions/population

Abbreviations: LD: linkage disequilibrium; AFR: African; AMR: Ad Mixed American; EAS: East Asian; EUR: European; SAS: South Asian.

Table 4: LD mean r^2 from 1000G[a] at various MAF thresholds

Population[b]	LD coverage (mean r^2)	
	MAF ≥ 5%	MAF ≥ 1%
AFR	0.47	0.31
AMR	0.71	0.53
EAS	0.77	0.64
EUR	0.74	0.59
SAS	0.72	0.56

a. Compared against Phase 3, version 5 of the 1000 Genomes Project (1000G). www.1000genomes.org. Accessed July 2016.
b. See www.1000genomes.org/category/frequently-asked-questions/population

Abbreviations: LD: linkage disequilibrium; MAF: minor allele frequency; AFR: African; AMR: Ad Mixed American; EAS: East Asian; EUR: European; SAS: South Asian.

Table 5: Marker information

Marker categories			No. of markers
Exonic markers[a]			268,631
Intronic markers[a]			152,454
Nonsense markers[b]			15,040
Missense markers[b]			219,228
Synonymous markers[b]			14,774
Mitochondrial markers[c]			369
Indels[c]			12,451
Sex chromosomes[c]	X 13,115	Y 2118	PAR/homologous 256

a. RefSeq - NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed September 2016.
b. Compared against the University of California, Santa Cruz (UCSC) Genome Browser. genome.ucsd.edu. Accessed August 2014.
c. NCBI Genome Reference Consortium, Version GRCh37. www.ncbi.nlm.nih.gov/grc/human. Accessed July 2016.

Abbreviations: indel: insertion/deletion; PAR: pseudoautosomal region.

Table 6: High-value content

Content	No. of markers	Research application/note
ADME core and extended genes[1]	10,177	Drug metabolism and excretion
APOE[2]	6	Cardiovascular disease, Alzheimer's disease, immunoregulation, and cognition
Blood phenotype genes[3]	798	Blood phenotypes
COSMIC[4] genes	364,900	Somatic mutations in cancer
GO[5] CVS genes	96,251	Cardiovascular conditions
Database of genomic variants[6]	432,491	Genomic structural variation
eQTLs[7]	3166	Genomic loci regulating mRNA expression levels
Fingerprint SNPs[8]	411	Human identification
HLA Genes[2]	306	Disease defense, transplant rejection, and autoimmune disorders
Extended MHC[a9]	5653	Disease defense, transplant rejection, and autoimmune disorders
KIR genes[2]	24	Autoimmune disorders and disease defense
Neanderthal SNPs[10]	607	Neanderthal ancestry and human population migration
NHGRI GWAS Catalog[11]	9405	Markers from published genome-wide association studies
RefSeq[12] 3' UTRs	19,749	3' untranslated regions of known genes
RefSeq 5' UTRs	15,055	5' untranslated regions of known genes
RefSeq All UTRs	33,856	All untranslated regions of known genes
RefSeq	391,037	All known genes
RefSeq +/- 10 kb	416,691	All known genes plus regulatory regions
RefSeq Promoters	13,200	2 kb upstream of all known genes to include promoter regions
RefSeq splice regions	7414	Variants at splice sites in all known genes

a. Extended MHC is a ~ 8 Mb region.

Abbreviations: ADME: absorption, distribution, metabolism, and excretion; APOE: apolipoprotein E; COSMIC: catalog of somatic mutations in cancer; GO CVS: gene ontology annotation of the cardiovascular system; eQTL: expression quantitative trait loci; HLA: human leukocyte antigen; KIR: killer cell immunoglobulin-like receptor; MHC: major histocompatibility complex; NHGRI: national human genome research institute; GWAS: genome-wide association study; UTR: untranslated region; RefSeq: reference sequence.

Ordering information

Infinium CoreExome-24 v1.3 Kit	Catalog no.
48 Samples	20024662
288 Samples	20024663
1152 Samples	20024664
Infinium CoreExome-24+ v1.3 Kit[a]	**Catalog no.**
48 Samples	20024665
288 Samples	20024666
1152 Samples	20024667

a. Enabled for additional custom content.

Learn more

To learn more about the Infinium CoreExome-24 v1.3 BeadChip and other Illumina genotyping products and services, visit www.illumina.com/genotyping

References

1. PharmaADME Gene List. www.pharmaadme.org. Accessed August 2014.
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3. NCBI Reference Sequence Blood Group Antigen Gene Mutation Database. www.ncbi.nlm.nih.gov/projects/gv/rbc/xslcgi.fcgi?cmd=bgmut/systems. Accessed July 2016.
4. Catalog of somatic mutations in cancer. cancer.sanger.uk/cosmic. Accessed July 2016.
5. Gene Ontology Consortium. www.geneontology.org. Accessed July 2016.
6. Database of Genomic Variants. dgv.tcag.ca/dgv/app/home. Accessed July 2016.
7. NCBI eQTL Database. www.ncbi.nlm.nih.gov/projects/gap/eqtl/index.cgi. Accessed July 2016.
8. The Allele Frequency Database. alfred.med.yale.edu/alfred/snpSets.asp. Accessed July 2016.
9. de Bakker PIW, McVean G, Sabeti PC, et al. A high-resolution HLA and SNP haplotype map for disease association studies in the extended human MHC. *Nat Genet*. 2006;38:1166–1172.
10. Neanderthal Genome Browser. neandertal.ensemblgenomes.org/index.html. Accessed July 2016.
11. NHGRI GWAS Catalog. www.ebi.ac.uk/gwas/docs/downloads. Accessed July 2016.
12. NCBI Reference Sequence Database. www.ncbi.nlm.nih.gov/refseq. Accessed July 2016.

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